

SEQUENCE LISTING

(1) GENERAL INFORMATION:

- (i) APPLICANT: Thomas, Winston J.
Drayna, Dennis T.
Feder, John N.
Gnirke, Andreas
Ruddy, David
Tsuchihashi, Zenta
Wolff, Roger K.
- (ii) TITLE OF INVENTION: Hereditary Hemochromatosis Gene
- (iii) NUMBER OF SEQUENCES: 44
- (iv) CORRESPONDENCE ADDRESS:
(A) ADDRESSEE: Townsend and Townsend and Crew LLP
(B) STREET: Two Embarcadero Center, Eighth Floor
(C) CITY: San Francisco
(D) STATE: California
(E) COUNTRY: USA
(F) ZIP: 94111-3834
- (v) COMPUTER READABLE FORM:
(A) MEDIUM TYPE: Floppy disk
(B) COMPUTER: IBM PC compatible
(C) OPERATING SYSTEM: PC-DOS/MS-DOS
(D) SOFTWARE: PatentIn Release #1.0, Version #1.30
- (vi) CURRENT APPLICATION DATA:
(A) APPLICATION NUMBER: US 08/652,265
(B) FILING DATE: 23-MAY-1996
(C) CLASSIFICATION:
- (viii) ATTORNEY/AGENT INFORMATION:
(A) NAME: Smith, William M.
(B) REGISTRATION NUMBER: 30,223
(C) REFERENCE/DOCKET NUMBER: 17957-000500
- (ix) TELECOMMUNICATION INFORMATION:
(A) TELEPHONE: (415) 576-0200
(B) TELEFAX: (415) 576-0300
- (2) INFORMATION FOR SEQ ID NO:1:
- (i) SEQUENCE CHARACTERISTICS:
(A) LENGTH: 10825 base pairs
(B) TYPE: nucleic acid
(C) STRANDEDNESS: single
(D) TOPOLOGY: linear
- (ii) MOLECULE TYPE: DNA (genomic)
- (ix) FEATURE:
(A) NAME/KEY: CDS
(B) LOCATION: join(361..436, 3762..4025, 4235..4510, 5606..5881,
6040..6153, 7107..7147)
(D) OTHER INFORMATION: /product= "Hereditary Hemochromatosis"

(HH) protein"
 /note= "Normal or wild-type (unaffected)
 Hereditary Hemochromatosis (HH) gene
 allele"

(ix) FEATURE:

(A) NAME/KEY: -
 (B) LOCATION: 140..7319
 (D) OTHER INFORMATION: /note= "start and stop positions for
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(ix) FEATURE:

(A) NAME/KEY: -
 (B) LOCATION: 3852..3891
 (D) OTHER INFORMATION: /note= "start and stop positions for
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(ix) FEATURE:

(A) NAME/KEY: -
 (B) LOCATION: 5507..6023
 (D) OTHER INFORMATION: /note= "start and stop positions for
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(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(3872, "c")
 (D) OTHER INFORMATION: /phenotype= "normal or wild-type
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 /label= 24d2

(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(3878, "a")
 (D) OTHER INFORMATION: /phenotype= "normal or wild-type
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 /label= 24d7

(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(5834, "g")
 (D) OTHER INFORMATION: /phenotype= "normal or wild-type
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 /label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:1:

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 ATTTTATAAA ACATTCTTCA CAACTCACA CACATTTAAA AACAAAACAC TGTCTCTAAA 10804
 ATCCCCAAT TTTTCATAAA C 10825

(2) INFORMATION FOR SEQ ID NO:2:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 348 amino acids
- (B) TYPE: amino acid
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: protein

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:2:

Met Gly Pro Arg Ala Arg Pro Ala Leu Leu Leu Met Leu Leu Gln
 1 5 10 15
 Thr Ala Val Leu Gln Gly Arg Leu Leu Arg Ser His Ser Leu His Tyr
 20 25 30
 Leu Phe Met Gly Ala Ser Glu Gln Asp Leu Gly Leu Ser Leu Phe Glu
 35 40 45
 Ala Leu Gly Tyr Val Asp Asp Gln Leu Phe Val Phe Tyr Asp His Glu

50					55					60					
Ser	Arg	Arg	Val	Glu	Pro	Arg	Thr	Pro	Trp	Val	Ser	Ser	Arg	Ile	Ser
65					70					75					80
Ser	Gln	Met	Trp	Leu	Gln	Leu	Ser	Gln	Ser	Leu	Lys	Gly	Trp	Asp	His
				85					90					95	
Met	Phe	Thr	Val	Asp	Phe	Trp	Thr	Ile	Met	Glu	Asn	His	Asn	His	Ser
			100					105					110		
Lys	Glu	Ser	His	Thr	Leu	Gln	Val	Ile	Leu	Gly	Cys	Glu	Met	Gln	Glu
		115					120					125			
Asp	Asn	Ser	Thr	Glu	Gly	Tyr	Trp	Lys	Tyr	Gly	Tyr	Asp	Gly	Gln	Asp
	130					135					140				
His	Leu	Glu	Phe	Cys	Pro	Asp	Thr	Leu	Asp	Trp	Arg	Ala	Ala	Glu	Pro
145					150					155					160
Arg	Ala	Trp	Pro	Thr	Lys	Leu	Glu	Trp	Glu	Arg	His	Lys	Ile	Arg	Ala
				165					170					175	
Arg	Gln	Asn	Arg	Ala	Tyr	Leu	Glu	Arg	Asp	Cys	Pro	Ala	Gln	Leu	Gln
			180					185					190		
Gln	Leu	Leu	Glu	Leu	Gly	Arg	Gly	Val	Leu	Asp	Gln	Gln	Val	Pro	Pro
		195					200					205			
Leu	Val	Lys	Val	Thr	His	His	Val	Thr	Ser	Ser	Val	Thr	Thr	Leu	Arg
	210					215					220				
Cys	Arg	Ala	Leu	Asn	Tyr	Pro	Gln	Asn	Ile	Thr	Met	Lys	Trp	Leu	
225				230					235					240	
Lys	Asp	Lys	Gln	Pro	Met	Asp	Ala	Lys	Glu	Phe	Glu	Pro	Lys	Asp	Val
				245					250					255	
Leu	Pro	Asn	Gly	Asp	Gly	Thr	Tyr	Gln	Gly	Trp	Ile	Thr	Leu	Ala	Val
			260					265					270		
Pro	Pro	Gly	Glu	Glu	Gln	Arg	Tyr	Thr	Cys	Gln	Val	Glu	His	Pro	Gly
		275					280					285			
Leu	Asp	Gln	Pro	Leu	Ile	Val	Ile	Trp	Glu	Pro	Ser	Pro	Ser	Gly	Thr
	290					295					300				
Leu	Val	Ile	Gly	Val	Ile	Ser	Gly	Ile	Ala	Val	Phe	Val	Val	Ile	Leu
305				310					315					320	
Phe	Ile	Gly	Ile	Leu	Phe	Ile	Ile	Leu	Arg	Lys	Arg	Gln	Gly	Ser	Arg
				325					330					335	
Gly	Ala	Met	Gly	His	Tyr	Val	Leu	Ala	Glu	Arg	Glu				
		340						345							

(2) INFORMATION FOR SEQ ID NO:3:

(i) SEQUENCE CHARACTERISTICS:

(A) LENGTH: 10825 base pairs

(B) TYPE: nucleic acid

- (C) STRANDEDNESS: single
(D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(ix) FEATURE:

- (A) NAME/KEY: CDS
(B) LOCATION: join(361..436, 3762..4025, 4235..4510, 5606..5881, 6040..6153, 7107..7147)
(D) OTHER INFORMATION: /product= "Hereditary Hemochromatosis (HH) protein containing the 24d1 mutation"
/note= "Hereditary Hemochromatosis (HH) gene 24d1 allele"

(ix) FEATURE:

- (A) NAME/KEY: -
(B) LOCATION: 140..7319
(D) OTHER INFORMATION: /note= "start and stop positions for 24d1 allele cDNA (SEQ ID NO:10)"

(ix) FEATURE:

- (A) NAME/KEY: -
(B) LOCATION: 3852..3891
(D) OTHER INFORMATION: /note= "start and stop positions for genomic sequence surrounding variant for 24d2(C) allele (SEQ ID NO:41)"

(ix) FEATURE:

- (A) NAME/KEY: -
(B) LOCATION: 5507..6023
(D) OTHER INFORMATION: /note= "start and stop positions for genomic sequence surrounding variant for 24d1(A) allele (SEQ ID NO:21)"

(ix) FEATURE:

- (A) NAME/KEY: allele
(B) LOCATION: replace(5834, "a")
(D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis (HH)"
/label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:3:

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TCAGGATTTA AAAACCAAGG GGGACACTGG ATCACCTAGT	GTTTCACAAG CAGGTACCTT	180
CTGCTGTAGG AGAGAGAGAA CTAAAGTTCT GAAAGACCTG	TTGCTTTTCA CCAGGAAGTT	240
TTACTGGGCA TCTCCTGAGC CTAGGCAATA GCTGTAGGGT	GACTTCTGGA GCCATCCCCG	300
TTTCCCCGCC CCCCAAAGA AGCGGAGATT TAACGGGGAC	GTGCGGCCAG AGCTGGGGAA	360
ATG GGC CCG CGA GCC AGG CCG GCG CTT CTC CTC	CTG ATG CTT TTG CAG	408
Met Gly Pro Arg Ala Arg Pro Ala Leu Leu Leu	Leu Met Leu Leu Gln	
1 5 10 15		

ACC GCG GTC CTG CAG GGG CGC TTG CTG C GTGAGTCCGA GGGCTGCGGG	456
Thr Ala Val Leu Gln Gly Arg Leu Leu	
20 25	
CGAACTAGGG GCGCGGCGGG GGTGGAAAAA TCGAACTAG CTTTTTCTTT GCGCTTGGGA	516
GTTTGCTAAC TTTGGAGGAC CTGCTCAACC CTATCCGCAA GCCCCTCTCC CTACTTTCTG	576
CGTCCAGACC CCGTGAGGGA GTGCCTACCA CTGAACTGCA GATAGGGGTC CCTCGCCCCA	636
GGACCTGCCC CCTCCCCCGG CTGTCCCGGC TCTGCGGAGT GACTTTTGGGA ACCGCCCACT	696
CCCTTCCCCC AACTAGAATG CTTTTAAATA AATCTCGTAG TTCCCTCACTT GAGCTGAGCT	756
AAGCCTGGGG CTCCTTGAAC CTGGAACCTCG GGTTTATTTT CAATGTCAGC TGTGCAGTTT	816
TTTCCCCAGT CATCTCCAAA CAGGAAGTTC TTCCCTGAGT GCTTGCCGAG AAGGCTGAGC	876
AAACCCACAG CAGGATCCGC ACGGGGTTTT CACCTCAGAA CGAATGCGTT GGGCGGTGGG	936
GGCGCGAAAG AGTGGCGTTG GGGATCTGAA TTCTTCACCA TTCCACCCAC TTTTGGTGAG	996
ACCTGGGGTG GAGGTCTCTA GGGTGGGAGG CTCCTGAGAG AGGCCTACCT CGGGCCTTTC	1056
CCCCTCTTG GCAATTGTTT TTTTGCCTGG AAAATTAAGT ATATGTTAGT TTTGAACGTT	1116
TGAACTGAAC AATTCTCTTT TCGGCTAGGC TTTATTGATT TGCAATGTGC TGTGTAATTA	1176
AGAGGCCTCT CTACAAAGTA CTGATAATGA ACATGTAAGC AATGCACTCA CTTCTAAGTT	1236
ACATTCATAT CTGATCTTAT TTGATTTTCA CTAGGCATAG GGAGGTAGGA GCTAATAATA	1296
CGTTTATTTT ACTAGAAGTT AACTGGAATT CAGATTATAT AACTCTTTTC AGGTTACAAA	1356
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GTTGACAGTG ATTTTGGCCCT GTAGTGTAGC ACAGTGTTC TGTGGGTCACA CGCCGGCCTC	1476
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GGCATGGCAC GGCCTGCTTC CTGGCAAATT TATTCAATGG TACTACTGGGC TTTGGTGGCA	1596
GAGCTCATGT CTCCACTTCA TAGCTATGAT TCTTAAACAT CACTACTGCAT TAGAGGTTGA	1656
ATAATAAAAT TTCATGTTGA GCAGAAATAT TCATTGTTTA CAAGTGTAAG TGAGTCCCAG	1716
CCATGTGTTG CACTGTTCAA GCCCAAGGG AGAGAGCAGG GAAACAAGTC TTTACCCTTT	1776
GATATTTTGC ATTCTAGTGG GAGAGATGAC AATAAGCAAA TGAGCAGAAA GATATACAAC	1836
ATCAGGAAAT CATGGGTGTT GTGAGAAGCA GAGAAGTCAG GGCAAGTCAC TCTGGGGCTG	1896
ACACTTGAGC AGAGACATGA AGGAAATAAG AATGATATTG ACTGGGAGCA GTATTTCCCA	1956
GGCAAACCTGA GTGGGCCTGG CAAGTTGGAT TAAAAAGCGG GTTTTCTCAG CACTACTCAT	2016
GTGTGTGTGT GTGGGGGGGG GGGGCGGCGT GGGGGTGGGA AGGGGGACTA CCATCTGCAT	2076
GTAGGATGTC TAGCAGTATC CTGTCCTCCC TACTCACTAG GTGCTAGGAG CACTCCCCCA	2136
GTCTTGACAA CCAAAAATGT CTCTAAACTT TGCCACATGT CACCTAGTAG ACAAACCTCT	2196

GGTTAAGAAG CTCGGGTTGA AAAAAATAAA CAAGTAGTGC TGGGGAGTAG AGGCCAAGAA 2256

GTAGGTAATG GGCTCAGAAG AGGAGCCACA AACAAGGTTG TGCAGGCGCC TGTAGGCTGT 2316

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GGCTGCTATG TGGAAAGCAG AATGAAGGGA GCAACAGTAA AAGCAGGGAG CCCAGCCAGG 2436

AAGCTGTTAC ACAGTCCAGG CAAGAGGTAG TGGAGTGGGC TGGGTGGGAA CAGAAAAGGG 2496

AGTGACAAAC CATTGTCTCC TGAATATATT CTGAAGGAAG TTGCTGAAGG ATTCTATGTT 2556

GTGTGAGAGA AAGAGAAGAA TTGGCTGGGT GTAGTAGCTC ATGCCAAGGA GGAGGCCAAG 2616

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TCTCTACAAA AAATACAAA ATTAGCTGGG TGTGGTGGCA TGCACCTGTG ATCCTAGCTA 2736

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AGTGGTGAGG CCACATAGGC AGTTCAGTGT AAGAATTCAG GACCAAGGCT GGGCACGGTG 3096

GCTCACTTCT GTAATCCCAG CACTTTGGTG GCTGAGGCAG GTAGATCATT TGAGGTCAGG 3156

AGTTTGAGAC AAGCTTGGCC AACATGGTGA AACCCCATGT CTAATAAAAA TACAAAAATT 3216

AGCCTGGTGT GGTGGCGCAC GCCTATAGTC CCAGGTTTTT AGGAGGCTTA GGTAGGAGAA 3276

TCCCTTGAAC CCAGGAGGTG CAGGTTGCAG TGAGCTGAGA TTGTGCCACT GCACTCCAGC 3336

CTGGGTGATA GAGTGAGACT CTGTCTCAAA AAAAAAAAAA AAAAAAAAAA AAAAACTGA 3396

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TACCATGGCT AGACACACCT TAACATTTTC TAGAATCCAC CAGCTTTAGT GGAGTCTGTC 3516

TAATCATGAG TATTGGAATA GGATCTGGGG GCAGTGAGGG GGTGGCAGCC ACGTGTGGCA 3576

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GTGGAGCCTC AACATCCTGC TCCCCTCCTA CTACACATGG TTAAGGCCTG TTGCTCTGTC 3756

TCCAG GT TCA CAC TCT CTG CAC TAC CTC TTC ATG GGT GCC TCA GAG 3802
Arg Ser His Ser Leu His Tyr Leu Phe Met Gly Ala Ser Glu
30 35

CAG GAC CTT GGT CTT TCC TTG TTT GAA GCT TTG GGC TAC GTG GAT GAC 3850
Gln Asp Leu Gly Leu Ser Leu Phe Glu Ala Leu Gly Tyr Val Asp Asp
40 45 50 55

CAG CTG TTC GTG TTC TAT GAT CAT GAG AGT CGC CGT GTG GAG CCC CGA 3898
Gln Leu Phe Val Phe Tyr Asp His Glu Ser Arg Arg Val Glu Pro Arg

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ACT CCA TGG GTT TCC AGT AGA ATT TCA AGC CAG ATG TGG CTG CAG CTG Thr Pro Trp Val Ser Ser Arg Ile Ser Ser Gln Met Trp Leu Gln Leu 75 80 85			3946
AGT CAG AGT CTG AAA GGG TGG GAT CAC ATG TTC ACT GTT GAC TTC TGG Ser Gln Ser Leu Lys Gly Trp Asp His Met Phe Thr Val Asp Phe Trp 90 95 100			3994
ACT ATT ATG GAA AAT CAC AAC CAC AGC AAG G GTATGTGGAG AGGGGGCCTC Thr Ile Met Glu Asn His Asn His Ser Lys 105 110			4045
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CAA GAA GAC AAC AGT ACC GAG GGC TAC TGG AAG TAC GGG TAT GAT GGG Gln Glu Asp Asn Ser Thr Glu Gly Tyr Trp Lys Tyr Gly Tyr Asp Gly 130 135 140			4320
CAG GAC CAC CTT GAA TTC TGC CCT GAC ACA CTG GAT TGG AGA GCA GCA Gln Asp His Leu Glu Phe Cys Pro Asp Thr Leu Asp Trp Arg Ala Ala 145 150 155			4368
GAA CCC AGG GCC TGG CCC ACC AAG CTG GAG TGG GAA AGG CAC AAG ATT Glu Pro Arg Ala Trp Pro Thr Lys Leu Glu Trp Glu Arg His Lys Ile 160 165 170			4416
CGG GCC AGG CAG AAC AGG GCC TAC CTG GAG AGG GAC TGC CCT GCA CAG Arg Ala Arg Gln Asn Arg Ala Tyr Leu Glu Arg Asp Cys Pro Ala Gln 175 180 185 190			4464
CTG CAG CAG TTG CTG GAG CTG GGG AGA GGT GTT TTG GAC CAA CAA G Leu Gln Gln Leu Leu Glu Leu Gly Arg Gly Val Leu Asp Gln Gln 195 200 205			4510
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ACAAGTCATG GGTTTAATTT CTTTCTCTCCA TGCATATGGC TCAAAGGGAA GTGTCTATGG			4750
CCCTTGCTTT TTATTTAACC AATAATCTTT TGTATATTTA TACCTGTAA AAATTCAGAA			4810
ATGTCAAGGC CGGGCACGGT GGCTCACCCC TGTAATCCCA GCACTTTGGG AGGCCGAGGC			4870
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TATCTATCAG AACAAAGAAC ATGGGTAACA GATATGTATA TTTACATGTG AGGAGAACAA 5410

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Val Pro Pro Leu Val Lys Val Thr His His Val Thr
210 215

TCT TCA GTG ACC ACT CTA CGG TGT CGG GCC TTG AAC TAC TAC CCC CAG 5688
Ser Ser Val Thr Thr Leu Arg Cys Arg Ala Leu Asn Tyr Tyr Pro Gln
220 225 230

AAC ATC ACC ATG AAG TGG CTG AAG GAT AAG CAG CCA ATG GAT GCC AAG 5736
Asn Ile Thr Met Lys Trp Leu Lys Asp Lys Gln Pro Met Asp Ala Lys
235 240 245

GAG TTC GAA CCT AAA GAC GTA TTG CCC AAT GGG GAT GGG ACC TAC CAG 5784
Glu Phe Glu Pro Lys Asp Val Leu Pro Asn Gly Asp Gly Thr Tyr Gln
250 255 260 265

GGC TGG ATA ACC TTG GCT GTA CCC CCT GGG GAA GAG CAG AGA TAT ACG 5832
Gly Trp Ile Thr Leu Ala Val Pro Pro Gly Glu Glu Gln Arg Tyr Thr
270 275 280

TAC CAG GTG GAG CAC CCA GGC CTG GAT CAG CCC CTC ATT GTG ATC TGG G 5881
Tyr Gln Val Glu His Pro Gly Leu Asp Gln Pro Leu Ile Val Ile Trp
285 290 295

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Glu Pro Ser Pro Ser
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GGC ACC CTA GTC ATT GGA GTC ATC AGT GGA ATT GCT GTT TTT GTC GTC 6101
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ATC TTG TTC ATT GGA ATT TTG TTC ATA ATA TTA AGG AAG AGG CAG GGT 6149
Ile Leu Phe Ile Gly Ile Leu Phe Ile Ile Leu Arg Lys Arg Gln Gly
320 325 330

TCA A GTGAGTAGGA ACAAGGGGGA AGTCTCTTAG TACCTCTGCC CCAGGGCACA 6203
Ser
335

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														345
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GGGTTTACC	ATGTTGGCCA	GGCTGGTCTC	GAACTCTCCT	GACCTCGTGA	TCCGCCTGCC	9304
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TCTTAATATA	TATATCCAGA	TGGCATGTGT	TTACTTTATG	TTACTACATG	CACTTGGCTG	9424
CATAAATGTG	GTACAAGCAT	TCTGTCTTGA	AGGGCAGGTG	CTTCAGGATA	CCATATACAG	9484
CTCAGAAGTT	TCTTCTTTAG	GCATTAAATT	TTAGCAAAGA	TATCTCATCT	CTTCTTTTAA	9544
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(2) INFORMATION FOR SEQ ID NO:4:

- (i) SEQUENCE CHARACTERISTICS:
- (A) LENGTH: 348 amino acids
 - (B) TYPE: amino acid
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: protein

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:4:

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 Leu Phe Met Gly Ala Ser Glu Gln Asp Leu Gly Leu Ser Leu Phe Glu
 35 40 45
 Ala Leu Gly Tyr Val Asp Asp Gln Leu Phe Val Phe Tyr Asp His Glu
 50 55 60
 Ser Arg Arg Val Glu Pro Arg Thr Pro Trp Val Ser Ser Arg Ile Ser
 65 70 75 80
 Ser Gln Met Trp Leu Gln Leu Ser Gln Ser Leu Lys Gly Trp Asp His

85										90					95				
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Lys	Glu	Ser	His	Thr	Leu	Gln	Val	Ile	Leu	Gly	Cys	Glu	Met	Gln	Glu				
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His	Leu	Glu	Phe	Cys	Pro	Asp	Thr	Leu	Asp	Trp	Arg	Ala	Ala	Glu	Pro				
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	210					215					220								
Cys	Arg	Ala	Leu	Asn	Tyr	Tyr	Pro	Gln	Asn	Ile	Thr	Met	Lys	Trp	Leu				
	225				230					235					240				
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Leu	Pro	Asn	Gly	Asp	Gly	Thr	Tyr	Gln	Gly	Trp	Ile	Thr	Leu	Ala	Val				
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Pro	Pro	Gly	Glu	Glu	Gln	Arg	Tyr	Thr	Tyr	Gln	Val	Glu	His	Pro	Gly				
		275					280					285							
Leu	Asp	Gln	Pro	Leu	Ile	Val	Ile	Trp	Glu	Pro	Ser	Pro	Ser	Gly	Thr				
	290					295					300								
Leu	Val	Ile	Gly	Val	Ile	Ser	Gly	Ile	Ala	Val	Phe	Val	Val	Ile	Leu				
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Phe	Ile	Gly	Ile	Leu	Phe	Ile	Ile	Leu	Arg	Lys	Arg	Gln	Gly	Ser	Arg				
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Gly	Ala	Met	Gly	His	Tyr	Val	Leu	Ala	Glu	Arg	Glu								
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(2) INFORMATION FOR SEQ ID NO:5:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 10825 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(ix) FEATURE:

- (A) NAME/KEY: CDS
- (B) LOCATION: join(361..436, 3762..4025, 4235..4510, 5606..5881, 6040..6153, 7107..7147)
- (D) OTHER INFORMATION: /product= "Hereditary Hemochromatosis (HH) protein containing the 24d2 mutation"
/note= "Hereditary Hemochromatosis (HH) gene 24d2 allele"

(ix) FEATURE:

- (A) NAME/KEY: -
- (B) LOCATION: 140..7319
- (D) OTHER INFORMATION: /note= "start and stop positions for 24d2 allele cDNA (SEQ ID NO:11)"

(ix) FEATURE:

- (A) NAME/KEY: -
- (B) LOCATION: 3852..3891
- (D) OTHER INFORMATION: /note= "start and stop positions for genomic sequence surrounding variant for 24d2(G) allele (SEQ ID NO:42)"

(ix) FEATURE:

- (A) NAME/KEY: -
- (B) LOCATION: 5507..6023
- (D) OTHER INFORMATION: /note= "start and stop positions for genomic sequence surrounding variant for 24d1(G) allele (SEQ ID NO:20)"

(ix) FEATURE:

- (A) NAME/KEY: allele
- (B) LOCATION: replace(3872, "g")
- (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis (HH)"
/label= 24d2

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:5:

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TTTCCCCGCC CCCCAAAGA AGCGGAGATT TAACGGGGAC GTGCGGCCAG AGCTGGGGAA      360
ATG GGC CCG CGA GCC AGG CCG GCG CTT CTC CTC CTG ATG CTT TTG CAG      408
Met Gly Pro Arg Ala Arg Pro Ala Leu Leu Leu Leu Met Leu Leu Gln
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          20          25
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ATCTCAAAAA	AATAAAAATA	AAAATAAAAA	AATGAAAAAA	AAAAGAAAGT	GAAGTATAGA	8464
GTATCTCATA	GTTTGTCACT	GATAGAAACA	GGTTTCAAAC	TCAGTCAATC	TGACCGTTTG	8524
ATACATCTCA	GACACCACTA	CATTCACTAG	TTTAGATGCC	TAGAATAAAT	AGAGAAGGAA	8584
GGAGATGGCT	CTTCTCTTGT	CTCATTGTGT	TTCTTCTGAG	TGAGCTTGAA	TCACATGAAG	8644
GGGAACAGCA	GAAAACAACC	AACTGATCCT	CAGCTGTCAT	GTTTCCTTTA	AAAGTCCCTG	8704
AAGGAAAGTC	CTGGAATGTG	ACTCCCTTGC	TCCTCTGTTG	CTCTCTTTGG	CATTCATTTT	8764
TTTGGACCCCT	ACGCAAGGAC	TGTAATTGGT	GGGGACAGCT	AGTGGCCCTG	CTGGGCTTCA	8824
CACACGGTGT	CCTCCCTAGG	CCAGTGCCTC	TGGAGTCAGA	ACTCTGGTGG	TATTTCCCTC	8884
AATGAAGTGG	AGTAAGCTCT	CTCATTTTGA	GATGGTATAA	TGGAAGCCAC	CAAGTGGCTT	8944
AGAGGATGCC	CAGGTCCTTC	CATGGAGCCA	CTGGGGTTCC	GGTGCACATT	AAAAAAAAAA	9004
TCTAACCAGG	ACATTCAAGG	ATTGCTAGAT	TCTGGGAAAT	CAGTTCACCA	TGTTCAAAAG	9064
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GCTCACTGTA	ACCTCTGCCT	CCCAGGTTCA	AGCGATTCTC	CTGTCTCAGC	CTCCCAAGTA	9184
GCTGGGATTA	CAGGCGTGCA	CCACCATGCC	CGGCTAATTT	TTGTATTTTT	AGTAGAGACA	9244
GGGTTTCACC	ATGTTGGCCA	GGCTGGTCTC	GAAGTCTCCT	GACCTCGTGA	TCCGCCTGCC	9304
TCGGCCTCCC	AAAGTGCTGA	GATTACAGGT	GTGAGCCACC	CTGCCCAGCC	GTCAAAGAG	9364
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CATAAATGTG	GTACAAGCAT	TCTGTCTTGA	AGGGCAGGTG	CTTCAGGATA	CCATATACAG	9484
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ACCATTTTCT	TTTTTTGTGG	TTAGAAAAGT	TATGTAGAAA	AAAGTAAATG	TGATTTACGC	9604
TCATTGTAGA	AAAGCTATAA	AATGAATACA	ATTAAAGCTG	TTATTTAATT	AGCCAGTGAA	9664
AAACTATTAA	CAACTTGTCT	ATTACCTGTT	AGTATTATTG	TTGCATTAAA	AATGCATATA	9724
CTTTAATAAA	TGTATATTGT	ATTGTATACT	GCATGATTTT	ATTGAAGTTC	TTGTTTCATCT	9784
TGTGTATATA	CTTAATCGCT	TTGTCAATTT	GGAGACATTT	ATTTTGCTTC	TAATTTCTTT	9844
ACATTTTGTC	TTACGGAATA	TTTTCAATCA	ACTGTGGTAG	CCGAATTAAT	CGTGTCTTCT	9904
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CTGAAAGCAT	ATGACAAATT	ATTTCTCTCT	TAATATCTTA	CTATACTGAA	AGCAGACTGC	10024

TATAAGGCTT CACTTACTCT TCTACCTCAT AAGGAATATG TTACAATTAA TTTATTAGGT 10084
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GACACCTGGT GGCCATAGGT AAATGTACCA CGGTGGTCCG GTGACCAGAG ATGCAGCGCT 10444
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AAAACAAACC ACTCTGATAA TCATTGAGTC AAGTACAGCA GGTGATTGAG GACTGCTGAG 10684
AGGTACAGGC CAAAATTCTT ATGTTGTATT ATAATAATGT CATCTTATAA TACTGTCAGT 10744
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ATCCCCAAAT TTTTCATAAA C 10825

(2) INFORMATION FOR SEQ ID NO:6:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 348 amino acids
- (B) TYPE: amino acid
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: protein

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:6:

Met Gly Pro Arg Ala Arg Pro Ala Leu Leu Leu Leu Met Leu Leu Gln
1 5 10 15
Thr Ala Val Leu Gln Gly Arg Leu Leu Arg Ser His Ser Leu His Tyr
20 25 30
Leu Phe Met Gly Ala Ser Glu Gln Asp Leu Gly Leu Ser Leu Phe Glu
35 40 45
Ala Leu Gly Tyr Val Asp Asp Gln Leu Phe Val Phe Tyr Asp Asp Glu
50 55 60
Ser Arg Arg Val Glu Pro Arg Thr Pro Trp Val Ser Ser Arg Ile Ser
65 70 75 80
Ser Gln Met Trp Leu Gln Leu Ser Gln Ser Leu Lys Gly Trp Asp His
85 90 95
Met Phe Thr Val Asp Phe Trp Thr Ile Met Glu Asn His Asn His Ser
100 105 110
Lys Glu Ser His Thr Leu Gln Val Ile Leu Gly Cys Glu Met Gln Glu

115					120					125					
Asp	Asn	Ser	Thr	Glu	Gly	Tyr	Trp	Lys	Tyr	Gly	Tyr	Asp	Gly	Gln	Asp
130						135					140				
His	Leu	Glu	Phe	Cys	Pro	Asp	Thr	Leu	Asp	Trp	Arg	Ala	Ala	Glu	Pro
145					150					155					160
Arg	Ala	Trp	Pro	Thr	Lys	Leu	Glu	Trp	Glu	Arg	His	Lys	Ile	Arg	Ala
				165					170					175	
Arg	Gln	Asn	Arg	Ala	Tyr	Leu	Glu	Arg	Asp	Cys	Pro	Ala	Gln	Leu	Gln
			180					185					190		
Gln	Leu	Leu	Glu	Leu	Gly	Arg	Gly	Val	Leu	Asp	Gln	Gln	Val	Pro	Pro
		195					200					205			
Leu	Val	Lys	Val	Thr	His	His	Val	Thr	Ser	Ser	Val	Thr	Thr	Leu	Arg
	210					215					220				
Cys	Arg	Ala	Leu	Asn	Tyr	Tyr	Pro	Gln	Asn	Ile	Thr	Met	Lys	Trp	Leu
225					230					235					240
Lys	Asp	Lys	Gln	Pro	Met	Asp	Ala	Lys	Glu	Phe	Glu	Pro	Lys	Asp	Val
				245					250					255	
Leu	Pro	Asn	Gly	Asp	Gly	Thr	Tyr	Gln	Gly	Trp	Ile	Thr	Leu	Ala	Val
			260					265					270		
Pro	Pro	Gly	Glu	Glu	Gln	Arg	Tyr	Thr	Cys	Gln	Val	Glu	His	Pro	Gly
		275					280					285			
Leu	Asp	Gln	Pro	Leu	Ile	Val	Ile	Trp	Glu	Pro	Ser	Pro	Ser	Gly	Thr
	290					295					300				
Leu	Val	Ile	Gly	Val	Ile	Ser	Gly	Ile	Ala	Val	Phe	Val	Val	Ile	Leu
305					310					315					320
Phe	Ile	Gly	Ile	Leu	Phe	Ile	Ile	Leu	Arg	Lys	Arg	Gln	Gly	Ser	Arg
			325					330						335	
Gly	Ala	Met	Gly	His	Tyr	Val	Leu	Ala	Glu	Arg	Glu				
			340					345							

(2) INFORMATION FOR SEQ ID NO:7:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 10825 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(ix) FEATURE:

- (A) NAME/KEY: CDS
- (B) LOCATION: join(361..436, 3762..4025, 4235..4510, 5606..5881, 6040..6153, 7107..7147)
- (D) OTHER INFORMATION: /product= "Hereditary Hemochromatosis (HH) protein containing both the 24d1

and 24d2 mutations"
 /note= "Hereditary Hemochromatosis (HH)
 gene containing a combination of both
 24d1 and 24d2 alleles"

(ix) FEATURE:

(A) NAME/KEY: -
 (B) LOCATION: 140..7319
 (D) OTHER INFORMATION: /note= "start and stop positions for
 cDNA containing a combination of both
 24d1 and 24d2 alleles
 (SEQ ID NO:12) "

(ix) FEATURE:

(A) NAME/KEY: -
 (B) LOCATION: 3852..3891
 (D) OTHER INFORMATION: /note= "start and stop positions for
 genomic sequence surrounding variant
 for 24d2(G) allele (SEQ ID NO:42) "

(ix) FEATURE:

(A) NAME/KEY: -
 (B) LOCATION: 5507..6023
 (D) OTHER INFORMATION: /note= "start and stop positions for
 genomic sequence surrounding variant
 for 24d1(A) allele (SEQ ID NO:21) "

(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(3872, "g")
 (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis
 (HH) "
 /label= 24d2

(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(5834, "a")
 (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis
 (HH) "
 /label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:7:

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TCAGGATTTA AAAACCAAGG GGGACACTGG ATCACCTAGT GTTTCACAAG CAGGTACCTT	180
CTGCTGTAGG AGAGAGAGAA CTAAAGTTCT GAAAGACCTG TTGCTTTTCA CCAGGAAGTT	240
TTACTGGGCA TCTCCTGAGC CTAGGCAATA GCTGTAGGGT GACTTCTGGA GCCATCCCCG	300
TTTCCCCGCC CCCCAAAGA AGCGGAGATT TAACGGGGAC GTGCGGCCAG AGCTGGGGAA	360
ATG GGC CCG CGA GCC AGG CCG GCG CTT CTC CTC CTG ATG CTT TTG CAG	408
Met Gly Pro Arg Ala Arg Pro Ala Leu Leu Leu Met Leu Leu Gln	
1 5 10 15	
ACC GCG GTC CTG CAG GGG CGC TTG CTG C GTGAGTCCGA GGGCTGCGGG	456
Thr Ala Val Leu Gln Gly Arg Leu Leu	

20

25

CGAACTAGGG	GCGCGGCGGG	GGTGGAAAAA	TCGAACTAG	CTTTTCTTT	GCGCTTGGGA	516
GTTTGCTAAC	TTTGGAGGAC	CTGCTCAACC	CTATCCGCA	GCCCCCTCC	CTACTTTCTG	576
CGTCCAGACC	CCGTGAGGGA	GTGCCTACCA	CTGAACTGCA	GATAGGGGTC	CCTCGCCCCA	636
GGACCTGCCC	CCTCCCCCGG	CTGTCCCGGC	TCTGCGGAGT	GACTTTTGGA	ACCGCCCACT	696
CCCTTCCCCC	AACTAGAATG	CTTTTAAATA	AATCTCGTAG	TTCCTCACTT	GAGCTGAGCT	756
AAGCCTGGGG	CTCCTTGAAC	CTGGAACTCG	GGTTTATTTT	CAATGTCAGC	TGTGCAGTTT	816
TTTCCCCAGT	CATCTCCAAA	CAGGAAGTTC	TTCCCTGAGT	GCTTGCCGAG	AAGGCTGAGC	876
AAACCCACAG	CAGGATCCGC	ACGGGGTTTC	CACCTCAGAA	CGAATGCGTT	GGGCGGTGGG	936
GGCGCGAAAAG	AGTGGCGTTG	GGGATCTGAA	TTCTTCACCA	TTCCACCCAC	TTTTGGTGAG	996
ACCTGGGGTG	GAGGTCTCTA	GGGTGGGAGG	CTCCTGAGAG	AGGCCTACCT	CGGGCCTTTC	1056
CCCACTCTTG	GCAATTGTTC	TTTTGCCTGG	AAAATTAAGT	ATATGTTAGT	TTTGAACGTT	1116
TGAACTGAAC	AATTCTCTTT	TCGGCTAGGC	TTTATTGATT	TGCAATGTGC	TGTGTAATTA	1176
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CGTTTATTTT	ACTAGAAGTT	AACTGGAATT	CAGATTATAT	AACTCTTTTC	AGGTTACAAA	1356
GAACATAAAAT	AATCTGGTTT	TCTGATGTTA	TTTCAAGTAC	TACAGCTGCT	TCTAATCTTA	1416
GTTGACAGTG	ATTTTGCCCT	GTAGTGTAGC	ACAGTGTCT	GTGGGTCACA	CGCCGGCCTC	1476
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GGCATGGCAC	GGCCTGCTTC	CTGGCAAATT	TATTCAATGG	TACACTGGGC	TTTGGTGGCA	1596
GAGCTCATGT	CTCCACTTCA	TAGCTATGAT	TCTTAAACAT	CACACTGCAT	TAGAGGTTGA	1656
ATAATAAAAT	TTCATGTTGA	GCAGAAATAT	TCATTGTTTA	CAAGTGTAAG	TGAGTCCCAG	1716
CCATGTGTTG	CACTGTTCAA	GCCCCAAGGG	AGAGAGCAGG	GAAACAAGTC	TTTACCCTTT	1776
GATATTTTGC	ATTCTAGTGG	GAGAGATGAC	AATAAGCAAA	TGAGCAGAAA	GATATACAAC	1836
ATCAGGAAAT	CATGGGTGTT	GTGAGAAGCA	GAGAAGTCAG	GGCAAGTCAC	TCTGGGGCTG	1896
ACACTTGAGC	AGAGACATGA	AGGAAATAAG	AATGATATTG	ACTGGGAGCA	GTATTTCCCA	1956
GGCAAACTGA	GTGGGCCTGG	CAAGTTGGAT	TAAAAAGCGG	GTTTTCTCAG	CACTACTCAT	2016
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GGTTAAGAAG	CTCGGGTTGA	AAAAAATAAA	CAAGTAGTGC	TGGGGAGTAG	AGGCCAAGAA	2256

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 GGTGTGAATT CTAGCCAAGG AGTAACAGTG ATCTGTCACA GGCTTTTAAA AGATTGCTCT 2376
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 30 35
 CAG GAC CTT GGT CTT TCC TTG TTT GAA GCT TTG GGC TAC GTG GAT GAC 3850
 Gln Asp Leu Gly Leu Ser Leu Phe Glu Ala Leu Gly Tyr Val Asp Asp 55
 40 45 50
 CAG CTG TTC GTG TTC TAT GAT GAT GAG AGT CGC CGT GTG GAG CCC CGA 3898
 Gln Leu Phe Val Phe Tyr Asp Asp Glu Ser Arg Arg Val Glu Pro Arg 70
 60 65 70

ACT CCA TGG GTT TCC AGT AGA ATT TCA AGC CAG ATG TGG CTG CAG CTG	3946
Thr Pro Trp Val Ser Ser Arg Ile Ser Ser Gln Met Trp Leu Gln Leu	
75 80 85	
AGT CAG AGT CTG AAA GGG TGG GAT CAC ATG TTC ACT GTT GAC TTC TGG	3994
Ser Gln Ser Leu Lys Gly Trp Asp His Met Phe Thr Val Asp Phe Trp	
90 95 100	
ACT ATT ATG GAA AAT CAC AAC CAC AGC AAG G GTATGTGGAG AGGGGGCCTC	4045
Thr Ile Met Glu Asn His Asn His Ser Lys	
105 110	
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TTTTTCCAG AG TCC CAC ACC CTG CAG GTC ATC CTG GGC TGT GAA ATG	4272
Glu Ser His Thr Leu Gln Val Ile Leu Gly Cys Glu Met	
115 120 125	
CAA GAA GAC AAC AGT ACC GAG GGC TAC TGG AAG TAC GGG TAT GAT GGG	4320
Gln Glu Asp Asn Ser Thr Glu Gly Tyr Trp Lys Tyr Gly Tyr Asp Gly	
130 135 140	
CAG GAC CAC CTT GAA TTC TGC CCT GAC ACA CTG GAT TGG AGA GCA GCA	4368
Gln Asp His Leu Glu Phe Cys Pro Asp Thr Leu Asp Trp Arg Ala Ala	
145 150 155	
GAA CCC AGG GCC TGG CCC ACC AAG CTG GAG TGG GAA AGG CAC AAG ATT	4416
Glu Pro Arg Ala Trp Pro Thr Lys Leu Glu Trp Glu Arg His Lys Ile	
160 165 170	
CGG GCC AGG CAG AAC AGG GCC TAC CTG GAG AGG GAC TGC CCT GCA CAG	4464
Arg Ala Arg Gln Asn Arg Ala Tyr Leu Glu Arg Asp Cys Pro Ala Gln	
175 180 185 190	
CTG CAG CAG TTG CTG GAG CTG GGG AGA GGT GTT TTG GAC CAA CAA G	4510
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195 200 205	
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CTCTTTTCTG	TCAAG	TG CCT CCT TTG	GTG AAG GTG ACA	CAT CAT GTG ACC			5640
		Val Pro Pro Leu	Val Lys Val Thr	His His Val Thr			
			210	215			
TCT TCA GTG ACC ACT CTA CGG TGT CGG GCC TTG AAC TAC TAC CCC CAG							5688
Ser Ser Val Thr Thr Leu Arg Cys Arg Ala Leu Asn Tyr Tyr Pro Gln							
	220		225		230		
AAC ATC ACC ATG AAG TGG CTG AAG GAT AAG CAG CCA ATG GAT GCC AAG							5736
Asn Ile Thr Met Lys Trp Leu Lys Asp Lys Gln Pro Met Asp Ala Lys							
	235		240		245		
GAG TTC GAA CCT AAA GAC GTA TTG CCC AAT GGG GAT GGG ACC TAC CAG							5784
Glu Phe Glu Pro Lys Asp Val Leu Pro Asn Gly Asp Gly Thr Tyr Gln							
	250		255		260		265
GGC TGG ATA ACC TTG GCT GTA CCC CCT GGG GAA GAG CAG AGA TAT ACG							5832
Gly Trp Ile Thr Leu Ala Val Pro Pro Gly Glu Glu Gln Arg Tyr Thr							
		270		275			280
TAC CAG GTG GAG CAC CCA GGC CTG GAT CAG CCC CTC ATT GTG ATC TGG G							5881
Tyr Gln Val Glu His Pro Gly Leu Asp Gln Pro Leu Ile Val Ile Trp							
	285		290		295		
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Gly Thr Leu Val Ile Gly Val Ile Ser Gly Ile Ala Val Phe Val Val							
	305		310		315		
ATC TTG TTC ATT GGA ATT TTG TTC ATA ATA TTA AGG AAG AGG CAG GGT							6149
Ile Leu Phe Ile Gly Ile Leu Phe Ile Ile Leu Arg Lys Arg Gln Gly							
	320		325		330		
TCA A GTGAGTAGGA	ACAAGGGGGA	AGTCTCTTAG	TACCTCTGCC	CCAGGGCACA			6203
Ser							
335							
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TAGAAATAAT	TACTGTACCT	TAACCCTGAG	TTTGCGTAGC	TATCACTCAC	CAATTATGCA	6743								
TTTCTACCCC	CTGAACATCT	GTGGTGTAGG	GAAAAGAGAA	TCAGAAAGAA	GCCAGCTCAT	6803								
ACAGAGTCCA	AGGGTCTTTT	GGGATATTGG	GTTATGATCA	CTGGGGTGTC	ATTGAAGGAT	6863								
CCTAAGAAAG	GAGGACCACG	ATCTCCCTTA	TATGGTGAAT	GTGTTGTTAA	GAAGTTAGAT	6923								
GAGAGGTGAG	GAGACCAGTT	AGAAAGCCAA	TAAGCATTTT	CAGATGAGAG	ATAATGGTTC	6983								
TTGAAATCCA	ATAGTGCCCA	GGTCTAAATT	GAGATGGGTG	AATGAGGAAA	ATAAGGAAGA	7043								
GAGAAGAGGC	AAGATGGTGC	CTAGGTTTGT	GATGCCTCTT	TCCTGGGTCT	CTTGTCTCCA	7103								
CAG	GA	GGA	GCC	ATG	GGG	CAC	TAC	GTC	TTA	GCT	GAA	CGT	GAG	7144
	Arg	Gly	Ala	Met	Gly	His	Tyr	Val	Leu	Ala	Glu	Arg	Glu	
					340									345
TGACACGCAG	CCTGCAGACT	CACTGTGGGA	AGGAGACAAA	ACTAGAGACT	CAAAGAGGGA	7204								
GTGCATTTAT	GAGCTCTTCA	TGTTTCAGGA	GAGAGTTGAA	CCTAAACATA	GAAATTGCCT	7264								
GACGAACTCC	TTGATTTTAG	CCTTCTCTGT	TCATTTCTCT	AAAAAGATTT	CCCCATTTAG	7324								
GTTTCTGAGT	TCCTGCATGC	CGGTGATCCC	TAGCTGTGAC	CTCTCCCCTG	GAAGTGTCTC	7384								
TCATGAACCT	CAAGCTGCAT	CTAGAGGCTT	CCTTCATTTT	CTCCGTCACC	TCAGAGACAT	7444								
ACACCTATGT	CATTTTCATTT	CCTATTTTTT	GAAGAGGACT	CCTTAAATTT	GGGGGACTTA	7504								
CATGATTCAT	TTTAACATCT	GAGAAAAGCT	TTGAACCCTG	GGACGTGGCT	AGTCATAACC	7564								
TTACCAGATT	TTTACACATG	TATCTATGCA	TTTTCTGGAC	CCGTTCAACT	TTTCCTTTGA	7624								
ATCCTCTCTC	TGTGTTACCC	AGTAACTCAT	CTGTCACCAA	GCCTTGGGGA	TTCTTCCATC	7684								
TGATTGTGAT	GTGAGTTGCA	CAGCTATGAA	GGCTGTACAC	TGCACGAATG	GAAGAGGCAC	7744								
CTGTCCCAGA	AAAAGCATCA	TGGCTATCTG	TGGGTAGTAT	GATGGGTGTT	TTTAGCAGGT	7804								
AGGAGGCAAA	TATCTTGAAA	GGGGTTGTGA	AGAGGTGTTT	TTTCTAATTG	GCATGAAGGT	7864								
GTCATACAGA	TTTGCAAAGT	TTAATGGTGC	CTTCATTTGG	GATGCTACTC	TAGTATTCCA	7924								
GACCTGAAGA	ATCACAATAA	TTTTCTACCT	GGTCTCTCCT	TGTTCTGATA	ATGAAAATTA	7984								
TGATAAGGAT	GATAAAAGCA	CTTACTTCGT	GTCCGACTCT	TCTGAGCACC	TACTTACATG	8044								

CATTACTGCA	TGCACTTCTT	ACAATAATTC	TATGAGATAG	GTACTATTAT	CCCCATTTCT	8104
TTTTTAAATG	AAGAAAGTGA	AGTAGGCCGG	GCACGGTGGC	TCACGCCTGT	AATCCCAGCA	8164
CTTTGGGAGG	CCAAAGCGGG	TGGATCACGA	GGTCAGGAGA	TCGAGACCAT	CCTGGCTAAC	8224
ATGGTGAAAC	CCCATCTCTA	ATAAAAATAC	AAAAAATTAG	CTGGGCGTGG	TGGCAGACGC	8284
CTGTAGTCCC	AGCTACTCGG	AAGGCTGAGG	CAGGAGAATG	GCATGAACCC	AGGAGGCAGA	8344
GCTTGCACTG	AGCCGAGTTT	GCGCCACTGC	ACTCCAGCCT	AGGTGACAGA	GTGAGACTCC	8404
ATCTCAAAAA	AATAAAAATA	AAAATAAAAA	AATGAAAAAA	AAAAGAAAGT	GAAGTATAGA	8464
GTATCTCATA	GTTTGTCACT	GATAGAAACA	GGTTTCAAAC	TCAGTCAATC	TGACCGTTTG	8524
ATACATCTCA	GACACCACTA	CATTCACTAG	TTTAGATGCC	TAGAATAAAT	AGAGAAGGAA	8584
GGAGATGGCT	CTTCTCTTGT	CTCATTGTGT	TTCTTCTGAG	TGAGCTTGAA	TCACATGAAG	8644
GGGAACAGCA	GAAAACAACC	AACTGATCCT	CAGCTGTCAT	GTTTCCTTTA	AAAGTCCCTG	8704
AAGGAAGGTC	CTGGAATGTG	ACTCCCTTGC	TCCTCTGTTG	CTCTCTTTGG	CATTCAATTC	8764
TTTGGACCTT	ACGCAAGGAC	TGTAATTGGT	GGGGACAGCT	AGTGGCCCTG	CTGGGCTTCA	8824
CACACGGTGT	CCTCCCTAGG	CCAGTGCCTC	TGGAGTCAGA	ACTCTGGTGG	TATTTCCCTC	8884
AATGAAGTGG	AGTAAGCTCT	CTCATTTTGA	GATGGTATAA	TGGAAGCCAC	CAAGTGGCTT	8944
AGAGGATGCC	CAGGTCCTTC	CATGGAGCCA	CTGGGGTTCC	GGTGCACATT	AAAAAAAAAA	9004
TCTAACCAGG	ACATTCAAGG	ATTGCTAGAT	TCTGGGAAAT	CAGTTCACCA	TGTTCAAAAG	9064
AGTCTTTTTT	TTTTTTTTTGA	GACTCTATTG	CCCAGGCTGG	AGTGCAATGG	CATGATCTCG	9124
GCTCACTGTA	ACCTCTGCCT	CCCAGGTTCA	AGCGATTCTC	CTGTCTCAGC	CTCCCAAGTA	9184
GCTGGGATTA	CAGGCGTGCA	CCACCATGCC	CGGCTAATTT	TTGTATTTTT	AGTAGAGACA	9244
GGGTTTCACC	ATGTTGGCCA	GGCTGGTCTC	GAACTCTCCT	GACCTCGTGA	TCCGCCTGCC	9304
TCGGCCTCCC	AAAGTGCTGA	GATTACAGGT	GTGAGCCACC	CTGCCCAGCC	GTCAAAGAG	9364
TCTTAATATA	TATATCCAGA	TGGCATGTGT	TTACTTTATG	TTACTACATG	CACTTGGCTG	9424
CATAAATGTG	GTACAAGCAT	TCTGTCTTGA	AGGGCAGGTG	CTTCAGGATA	CCATATACAG	9484
CTCAGAAGTT	TCTTCTTTAG	GCATTAAATT	TTAGCAAAGA	TATCTCATCT	CTTCTTTTAA	9544
ACCATTTTCT	TTTTTTGTGG	TTAGAAAAGT	TATGTAGAAA	AAAGTAAATG	TGATTTACGC	9604
TCATTGTAGA	AAAGCTATAA	AATGAATACA	ATTAAAGCTG	TTATTTAATT	AGCCAGTGAA	9664
AAACTATTAA	CAACTTGTCT	ATTACCTGTT	AGTATTATTG	TTGCATTAAA	AATGCATATA	9724
CTTTAATAAA	TGTATATTGT	ATTGTATACT	GCATGATTTT	ATTGAAGTTC	TTGTTCATCT	9784
TGTGTATATA	CTTAATCGCT	TTGTCATTTT	GGAGACATTT	ATTTTGCTTC	TAATTTCTTT	9844
ACATTTTGTC	TTACGGAATA	TTTTCAATCA	ACTGTGGTAG	CCGAATTAAT	CGTGTTTCTT	9904

CACTCTAGGG ACATTGTCGT CTAAGTTGTA AGACATTGGT TATTTTACCA GCAAACCATT 9964
 CTGAAAGCAT ATGACAAATT ATTTCTCTCT TAATATCTTA CTATACTGAA AGCAGACTGC 10024
 TATAAGGCTT CACTTACTCT TCTACCTCAT AAGGAATATG TTACAATTAA TTTATTAGGT 10084
 AAGCATTTGT TTTATATTGG TTTTATTTCA CCTGGGCTGA GATTTCAGA AACACCCCAG 10144
 TCTTCACAGT AACACATTTT ACTAACACAT TTAATAACA TCAGCAACTG TGGCCTGTTA 10204
 ATTTTTTTTAA TAGAAATTTT AAGTCCTCAT TTTCTTTTCG TGTTTTTTTAA GCTTAATTTT 10264
 TCTGGCTTTT TTCATAAATT CTTAAGGTCA ACTACATTTG AAAAATCAAA GACCTGCATT 10324
 TTAAATTCTT ATTCACCTCT GGCAAAACCA TTCACAAACC ATGGTAGTAA AGAGAAGGGT 10384
 GACACCTGGT GGCCATAGGT AAATGTACCA CGGTGGTCCG GTGACCAGAG ATGCAGCGCT 10444
 GAGGGTTTTT CTGAAGGTAA AGGAATAAAG AATGGGTGGA GGGGCGTGCA CTGGAAATCA 10504
 CTTGTAGAGA AAAGCCCCTG AAAATTTGAG AAAACAAACA AGAACTACT TACCAGCTAT 10564
 TTGAATTGCT GGAATCACAG GCCATTGCTG AGCTGCCTGA ACTGGGAACA CAACAGAAGG 10624
 AAAACAAACC ACTCTGATAA TCATTGAGTC AAGTACAGCA GGTGATTGAG GACTGCTGAG 10684
 AGGTACAGGC CAAAATTCTT ATGTTGTATT ATAATAATGT CATCTTATAA TACTGTCAGT 10744
 ATTTTATAAA ACATTCTTCA CAACTCACA CACATTTAAA AACAAAACAC TGTCTCTAAA 10804
 ATCCCCAAAT TTTTCATAAA C 10825

(2) INFORMATION FOR SEQ ID NO:8:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 348 amino acids
- (B) TYPE: amino acid
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: protein

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:8:

Met Gly Pro Arg Ala Arg Pro Ala Leu Leu Leu Met Leu Leu Gln
 1 5 10 15
 Thr Ala Val Leu Gln Gly Arg Leu Leu Arg Ser His Ser Leu His Tyr
 20 25 30
 Leu Phe Met Gly Ala Ser Glu Gln Asp Leu Gly Leu Ser Leu Phe Glu
 35 40 45
 Ala Leu Gly Tyr Val Asp Asp Gln Leu Phe Val Phe Tyr Asp Asp Glu
 50 55 60
 Ser Arg Arg Val Glu Pro Arg Thr Pro Trp Val Ser Ser Arg Ile Ser
 65 70 75 80
 Ser Gln Met Trp Leu Gln Leu Ser Gln Ser Leu Lys Gly Trp Asp His
 85 90 95

Met Phe Thr Val Asp Phe Trp Thr Ile Met Glu Asn His Asn His Ser
 100 105 110
 Lys Glu Ser His Thr Leu Gln Val Ile Leu Gly Cys Glu Met Gln Glu
 115 120 125
 Asp Asn Ser Thr Glu Gly Tyr Trp Lys Tyr Gly Tyr Asp Gly Gln Asp
 130 135 140
 His Leu Glu Phe Cys Pro Asp Thr Leu Asp Trp Arg Ala Ala Glu Pro
 145 150 155 160
 Arg Ala Trp Pro Thr Lys Leu Glu Trp Glu Arg His Lys Ile Arg Ala
 165 170 175
 Arg Gln Asn Arg Ala Tyr Leu Glu Arg Asp Cys Pro Ala Gln Leu Gln
 180 185 190
 Gln Leu Leu Glu Leu Gly Arg Gly Val Leu Asp Gln Gln Val Pro Pro
 195 200 205
 Leu Val Lys Val Thr His His Val Thr Ser Ser Val Thr Thr Leu Arg
 210 215 220
 Cys Arg Ala Leu Asn Tyr Tyr Pro Gln Asn Ile Thr Met Lys Trp Leu
 225 230 235 240
 Lys Asp Lys Gln Pro Met Asp Ala Lys Glu Phe Glu Pro Lys Asp Val
 245 250 255
 Leu Pro Asn Gly Asp Gly Thr Tyr Gln Gly Trp Ile Thr Leu Ala Val
 260 265 270
 Pro Pro Gly Glu Glu Gln Arg Tyr Thr Tyr Gln Val Glu His Pro Gly
 275 280 285
 Leu Asp Gln Pro Leu Ile Val Ile Trp Glu Pro Ser Pro Ser Gly Thr
 290 295 300
 Leu Val Ile Gly Val Ile Ser Gly Ile Ala Val Phe Val Val Ile Leu
 305 310 315 320
 Phe Ile Gly Ile Leu Phe Ile Ile Leu Arg Lys Arg Gln Gly Ser Arg
 325 330 335
 Gly Ala Met Gly His Tyr Val Leu Ala Glu Arg Glu
 340 345

(2) INFORMATION FOR SEQ ID NO:9:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 1440 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: cDNA

(ix) FEATURE:

- (A) NAME/KEY: CDS

(B) LOCATION: 222..1268

(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(408, "c")
 (D) OTHER INFORMATION: /phenotype= "normal or wild-type
 (unaffected)"
 /label= 24d2

(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(414, "a")
 (D) OTHER INFORMATION: /phenotype= "normal or wild-type
 (unaffected)"
 /label= 24d7

(ix) FEATURE:

(A) NAME/KEY: allele
 (B) LOCATION: replace(1066, "g")
 (D) OTHER INFORMATION: /phenotype= "normal or wild-type
 (unaffected)"
 /label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:9:

GGGGACACTG GATCACCTAG TGTTTCACAA GCAGGTACCT TCTGCTGTAG GAGAGAGAGA	60
ACTAAAGTTC TGAAAGACCT GTTGCTTTTC ACCAGGAAGT TTTACTGGGC ATCTCCTGAG	120
CCTAGGCAAT AGCTGTAGGG TGACTTCTGG AGCCATCCCC GTTTCCTCCG CCCCCAAAAG	180
AAGCGGAGAT TTAACGGGGA CGTGCGGCCA GAGCTGGGGA A ATG GGC CCG CGA	233
Met Gly Pro Arg	
1	
GCC AGG CCG GCG CTT CTC CTC CTG ATG CTT TTG CAG ACC GCG GTC CTG	281
Ala Arg Pro Ala Leu Leu Leu Leu Met Leu Leu Gln Thr Ala Val Leu	
5 10 15 20	
CAG GGG CGC TTG CTG CGT TCA CAC TCT CTG CAC TAC CTC TTC ATG GGT	329
Gln Gly Arg Leu Leu Arg Ser His Ser Leu His Tyr Leu Phe Met Gly	
25 30 35	
GCC TCA GAG CAG GAC CTT GGT CTT TCC TTG TTT GAA GCT TTG GGC TAC	377
Ala Ser Glu Gln Asp Leu Gly Leu Ser Leu Phe Glu Ala Leu Gly Tyr	
40 45 50	
GTG GAT GAC CAG CTG TTC GTG TTC TAT GAT CAT GAG AGT CGC CGT GTG	425
Val Asp Asp Gln Leu Phe Val Phe Tyr Asp His Glu Ser Arg Arg Val	
55 60 65	
GAG CCC CGA ACT CCA TGG GTT TCC AGT AGA ATT TCA AGC CAG ATG TGG	473
Glu Pro Arg Thr Pro Trp Val Ser Ser Arg Ile Ser Ser Gln Met Trp	
70 75 80	
CTG CAG CTG AGT CAG AGT CTG AAA GGG TGG GAT CAC ATG TTC ACT GTT	521
Leu Gln Leu Ser Gln Ser Leu Lys Gly Trp Asp His Met Phe Thr Val	
85 90 95 100	
GAC TTC TGG ACT ATT ATG GAA AAT CAC AAC CAC AGC AAG GAG TCC CAC	569

Asp	Phe	Trp	Thr	Ile 105	Met	Glu	Asn	His	Asn 110	His	Ser	Lys	Glu	Ser 115	His	
ACC Thr	CTG Leu	CAG Gln	GTC Val	ATC Ile 120	CTG Leu	GGC Gly	TGT Cys	GAA Glu 125	ATG Met	CAA Gln	GAA Glu	GAC Asp	AAC Asn 130	AGT Ser	ACC Thr	617
GAG Glu	GGC Gly	TAC Tyr 135	TGG Trp	AAG Lys	TAC Tyr	GGG Gly	TAT Tyr 140	GAT Asp	GGG Gly	CAG Gln	GAC Asp	CAC His 145	CTT Leu	GAA Glu	TTC Phe	665
TGC Cys	CCT Pro 150	GAC Asp	ACA Thr	CTG Leu	GAT Asp	TGG Trp 155	AGA Arg	GCA Ala	GCA Ala	GAA Glu	CCC Pro 160	AGG Arg	GCC Ala	TGG Trp	CCC Pro	713
ACC Thr 165	AAG Lys	CTG Leu	GAG Glu	TGG Trp 170	GAA Glu 170	AGG Arg	CAC His	AAG Lys	ATT Ile 175	CGG Arg 175	GCC Ala	AGG Arg	CAG Gln	AAC Asn 180	AGG Arg 180	761
GCC Ala	TAC Tyr	CTG Leu	GAG Glu	AGG Arg 185	GAC Asp	TGC Cys	CCT Pro	GCA Ala 190	CAG Gln 190	CTG Leu	CAG Gln	CAG Gln	TTG Leu 195	CTG Leu 195	GAG Glu 195	809
CTG Leu	GGG Gly	AGA Arg 200	GGT Gly 200	GTT Val	TTG Leu	GAC Asp	CAA Gln 205	CAA Gln 205	GTG Val	CCT Pro	CCT Pro	TTG Leu 210	GTG Val 210	AAG Lys	GTG Val	857
ACA Thr	CAT His 215	CAT His 215	GTG Val	ACC Thr	TCT Ser	TCA Ser 220	GTG Val 220	ACC Thr	ACT Thr	CTA Leu	CGG Arg 225	TGT Cys 225	CGG Arg	GCC Ala	TTG Leu	905
AAC Asn 230	TAC Tyr 230	TAC Tyr	CCC Pro	CAG Gln	AAC Asn	ATC Ile 235	ACC Thr 235	ATG Met	AAG Lys	TGG Trp 240	CTG Leu 240	AAG Lys	GAT Asp	AAG Lys	CAG Gln	953
CCA Pro 245	ATG Met	GAT Asp	GCC Ala	AAG Lys 250	GAG Glu 250	TTC Phe	GAA Glu	CCT Pro	AAA Lys	GAC Asp 255	GTA Val	TTG Leu	CCC Pro	AAT Asn	GGG Gly 260	1001
GAT Asp	GGG Gly	ACC Thr	TAC Tyr 265	CAG Gln 265	GGC Gly	TGG Trp	ATA Ile	ACC Thr 270	TTG Leu 270	GCT Ala	GTA Val	CCC Pro	CCT Pro	GGG Gly 275	GAA Glu 275	1049
GAG Glu	CAG Gln	AGA Arg 280	TAT Tyr 280	ACG Thr	TGC Cys	CAG Gln	GTG Val 285	GAG Glu 285	CAC His	CCA Pro	GGC Gly	CTG Leu 290	GAT Asp 290	CAG Gln	CCC Pro	1097
CTC Leu	ATT Ile 295	GTG Val 295	ATC Ile 295	TGG Trp	GAG Glu	CCC Pro	TCA Ser 300	CCG Pro	TCT Ser	GGC Gly	ACC Thr 305	CTA Leu 305	GTC Val 305	ATT Ile	GGA Gly	1145
GTC Val	ATC Ile 310	AGT Ser	GGA Gly	ATT Ile	GCT Ala	GTT Val 315	TTT Phe	GTC Val 315	GTC Val	ATC Ile 320	TTG Leu 320	TTC Phe	ATT Ile	GGA Gly	ATT Ile	1193
TTG Leu 325	TTC Phe	ATA Ile	ATA Ile	TTA Leu	AGG Arg 330	AAG Lys	AGG Arg	CAG Gln	GGT Gly	TCA Ser 335	AGA Arg	GGA Gly	GCC Ala	ATG Met	GGG Gly 340	1241
CAC His	TAC Tyr	GTC Val	TTA Leu	GCT Ala 345	GAA Glu	CGT Arg	GAG Glu	TGACACGCAG	GAG Glu	TGACACGCAG	CCTGCAGACT	CCTGCAGACT	CACTGTGGGA	CACTGTGGGA	CACTGTGGGA	1295

AGGAGACAAA ACTAGAGACT CAAAGAGGGA GTGCATTTAT GAGCTCTTCA TGTTTCAGGA 1355
 GAGAGTTGAA CCTAAACATA GAAATTGCCT GACGAACTCC TTGATTTTAG CTTTCTCTGT 1415
 TCATTTCTC AAAAAAGATTT CCCCCA 1440

(2) INFORMATION FOR SEQ ID NO:10:

- (i) SEQUENCE CHARACTERISTICS:
 (A) LENGTH: 1440 base pairs
 (B) TYPE: nucleic acid
 (C) STRANDEDNESS: single
 (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: cDNA

(ix) FEATURE:

- (A) NAME/KEY: CDS
 (B) LOCATION: 222..1268

(ix) FEATURE:

- (A) NAME/KEY: allele
 (B) LOCATION: replace(1066, "a")
 (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis (HH)"
 /label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:10:

GGGGACACTG GATCACCTAG TGTTTCACAA GCAGGTACCT TCTGCTGTAG GAGAGAGAGA 60
 ACTAAAGTTC TGAAAGACCT GTTGCTTTTC ACCAGGAAGT TTTACTGGGC ATCTCCTGAG 120
 CCTAGGCAAT AGCTGTAGGG TGACTTCTGG AGCCATCCCC GTTTCCCCGC CCCCCAAAAG 180
 AAGCGGAGAT TTAACGGGGA CGTGCGGCCA GAGCTGGGGA A ATG GGC CCG CGA 233
 Met Gly Pro Arg
 1
 GCC AGG CCG GCG CTT CTC CTC CTG ATG CTT TTG CAG ACC GCG GTC CTG 281
 Ala Arg Pro Ala Leu Leu Leu Met Leu Leu Gln Thr Ala Val Leu
 5 10 15 20
 CAG GGG CGC TTG CTG CGT TCA CAC TCT CTG CAC TAC CTC TTC ATG GGT 329
 Gln Gly Arg Leu Leu Arg Ser His Ser Leu His Tyr Leu Phe Met Gly
 25 30 35
 GCC TCA GAG CAG GAC CTT GGT CTT TCC TTG TTT GAA GCT TTG GGC TAC 377
 Ala Ser Glu Gln Asp Leu Gly Leu Ser Leu Phe Glu Ala Leu Gly Tyr
 40 45 50
 GTG GAT GAC CAG CTG TTC GTG TTC TAT GAT CAT GAG AGT CGC CGT GTG 425
 Val Asp Asp Gln Leu Phe Val Phe Tyr Asp His Glu Ser Arg Arg Val
 55 60 65
 GAG CCC CGA ACT CCA TGG GTT TCC AGT AGA ATT TCA AGC CAG ATG TGG 473
 Glu Pro Arg Thr Pro Trp Val Ser Ser Arg Ile Ser Ser Gln Met Trp
 70 75 80

CTG Leu 85	CAG Gln	CTG Leu	AGT Ser	CAG Gln	AGT Ser	CTG Leu	AAA Lys	GGG Gly	TGG Trp	GAT Asp	CAC His	ATG Met	TTC Phe	ACT Thr	GTT Val	521
GAC Asp	TTC Phe	TGG Trp	ACT Thr	ATT Ile	ATG Met	GAA Glu	AAT Asn	CAC His	AAC Asn	CAC His	AGC Ser	AAG Lys	GAG Glu	TCC Ser	CAC His	569
ACC Thr	CTG Leu	CAG Gln	GTC Val	ATC Ile	CTG Leu	GGC Gly	TGT Cys	GAA Glu	ATG Met	CAA Gln	GAA Glu	GAC Asp	AAC Asn	AGT Ser	ACC Thr	617
GAG Glu	GGC Gly	TAC Tyr	TGG Trp	AAG Lys	TAC Tyr	GGG Gly	TAT Tyr	GAT Asp	GGG Gly	CAG Gln	GAC Asp	CAC His	CTT Leu	GAA Glu	TTC Phe	665
TGC Cys	CCT Pro	GAC Asp	ACA Thr	CTG Leu	GAT Asp	TGG Trp	AGA Arg	GCA Ala	GCA Ala	GAA Glu	CCC Pro	AGG Arg	GCC Ala	TGG Trp	CCC Pro	713
ACC Thr	AAG Lys	CTG Leu	GAG Glu	TGG Trp	GAA Glu	AGG Arg	CAC His	AAG Lys	ATT Ile	CGG Arg	GCC Ala	AGG Arg	CAG Gln	AAC Asn	AGG Arg	761
GCC Ala	TAC Tyr	CTG Leu	GAG Glu	AGG Arg	GAC Asp	TGC Cys	CCT Pro	GCA Ala	CAG Gln	CTG Leu	CAG Gln	CAG Gln	TTG Leu	CTG Leu	GAG Glu	809
CTG Leu	GGG Gly	AGA Arg	GGT Gly	GTT Val	TTG Leu	GAC Asp	CAA Gln	CAA Gln	GTG Val	CCT Pro	CCT Pro	TTG Leu	GTG Val	AAG Lys	GTG Val	857
ACA Thr	CAT His	CAT His	GTG Val	ACC Thr	TCT Ser	TCA Ser	GTG Val	ACC Thr	ACT Thr	CTA Leu	CGG Arg	TGT Cys	CGG Arg	GCC Ala	TTG Leu	905
AAC Asn	TAC Tyr	TAC Tyr	CCC Pro	CAG Gln	AAC Asn	ATC Ile	ACC Thr	ATG Met	AAG Lys	TGG Trp	CTG Leu	AAG Lys	GAT Asp	AAG Lys	CAG Gln	953
CCA Pro	ATG Met	GAT Asp	GCC Ala	AAG Lys	GAG Glu	TTC Phe	GAA Glu	CCT Pro	AAA Lys	GAC Asp	GTA Val	TTG Leu	CCC Pro	AAT Asn	GGG Gly	1001
GAT Asp	GGG Gly	ACC Thr	TAC Tyr	CAG Gln	GGC Gly	TGG Trp	ATA Ile	ACC Thr	TTG Leu	GCT Ala	GTA Val	CCC Pro	CCT Pro	GGG Gly	GAA Glu	1049
GAG Glu	CAG Gln	AGA Arg	TAT Tyr	ACG Thr	TAC Tyr	CAG Gln	GTG Val	GAG Glu	CAC His	CCA Pro	GGC Gly	CTG Leu	GAT Asp	CAG Gln	CCC Pro	1097
CTC Leu	ATT Ile	GTG Val	ATC Ile	TGG Trp	GAG Glu	CCC Pro	TCA Ser	CCG Pro	TCT Ser	GGC Gly	ACC Thr	CTA Leu	GTC Val	ATT Ile	GGA Gly	1145
GTC Val	ATC Ile	AGT Ser	GGA Gly	ATT Ile	GCT Ala	GTT Val	TTT Phe	GTC Val	GTC Val	ATC Ile	TTG Leu	TTC Phe	ATT Ile	GGA Gly	ATT Ile	1193
TTG Leu	TTC Phe	ATA Ile	ATA Ile	TTA Leu	AGG Arg	AAG Lys	AGG Arg	CAG Gln	GGT Gly	TCA Ser	AGA Arg	GGA Gly	GCC Ala	ATG Met	GGG Gly	1241

325	330	335	340	
CAC TAC GTC TTA GCT GAA CGT GAG	TGACACGCAG	CCTGCAGACT	CACTGTGGGA	1295
His Tyr Val Leu Ala Glu Arg Glu				
	345			
AGGAGACAAA	ACTAGAGACT	CAAAGAGGGA	GTGCATTTAT	GAGCTCTTCA
				TGTTTCAGGA
				1355
GAGAGTTGAA	CCTAAACATA	GAAATTGCCT	GACGAACTCC	TTGATTTTAG
				CCTTCTCTGT
				1415
TCATTTCTCTC	AAAAAGATTT	CCCCA		1440

(2) INFORMATION FOR SEQ ID NO:11:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 1440 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: cDNA

(ix) FEATURE:

- (A) NAME/KEY: CDS
- (B) LOCATION: 222..1268

(ix) FEATURE:

- (A) NAME/KEY: allele
- (B) LOCATION: replace(408, "g")
- (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis (HH)"
/label= 24d2

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:11:

GGGGACACTG	GATCACCTAG	TGTTTCACAA	GCAGGTACCT	TCTGCTGTAG	GAGAGAGAGA	60
ACTAAAGTTC	TGAAAGACCT	GTTGCTTTTC	ACCAGGAAGT	TTTACTGGGC	ATCTCCTGAG	120
CCTAGGCAAT	AGCTGTAGGG	TGACTTCTGG	AGCCATCCCC	GTTTCCCCGC	CCCCCAAAG	180
AAGCGGAGAT	TTAACGGGGA	CGTGCGGCCA	GAGCTGGGGA	A ATG GGC CCG CGA		233
				Met Gly Pro Arg		
				1		
GCC AGG CCG GCG CTT CTC CTC CTG ATG CTT TTG CAG ACC GCG GTC CTG						281
Ala Arg Pro Ala Leu Leu Leu Met Leu Leu Gln Thr Ala Val Leu						
5	10		15		20	
CAG GGG CGC TTG CTG CGT TCA CAC TCT CTG CAC TAC CTC TTC ATG GGT						329
Gln Gly Arg Leu Leu Arg Ser His Ser Leu His Tyr Leu Phe Met Gly						
	25		30		35	
GCC TCA GAG CAG GAC CTT GGT CTT TCC TTG TTT GAA GCT TTG GGC TAC						377
Ala Ser Glu Gln Asp Leu Gly Leu Ser Leu Phe Glu Ala Leu Gly Tyr						
	40		45		50	
GTG GAT GAC CAG CTG TTC GTG TTC TAT GAT GAT GAG AGT CGC CGT GTG						425

Val	Asp	Asp	Gln	Leu	Phe	Val	Phe	Tyr	Asp	Asp	Glu	Ser	Arg	Arg	Val	
		55					60					65				
GAG	CCC	CGA	ACT	CCA	TGG	GTT	TCC	AGT	AGA	ATT	TCA	AGC	CAG	ATG	TGG	473
Glu	Pro	Arg	Thr	Pro	Trp	Val	Ser	Ser	Arg	Ile	Ser	Ser	Gln	Met	Trp	
	70					75					80					
CTG	CAG	CTG	AGT	CAG	AGT	CTG	AAA	GGG	TGG	GAT	CAC	ATG	TTC	ACT	GTT	521
Leu	Gln	Leu	Ser	Gln	Ser	Leu	Lys	Gly	Trp	Asp	His	Met	Phe	Thr	Val	
	85				90					95					100	
GAC	TTC	TGG	ACT	ATT	ATG	GAA	AAT	CAC	AAC	CAC	AGC	AAG	GAG	TCC	CAC	569
Asp	Phe	Trp	Thr	Ile	Met	Glu	Asn	His	Asn	His	Ser	Lys	Glu	Ser	His	
				105					110					115		
ACC	CTG	CAG	GTC	ATC	CTG	GGC	TGT	GAA	ATG	CAA	GAA	GAC	AAC	AGT	ACC	617
Thr	Leu	Gln	Val	Ile	Leu	Gly	Cys	Glu	Met	Gln	Glu	Asp	Asn	Ser	Thr	
			120					125					130			
GAG	GGC	TAC	TGG	AAG	TAC	GGG	TAT	GAT	GGG	CAG	GAC	CAC	CTT	GAA	TTC	665
Glu	Gly	Tyr	Trp	Lys	Tyr	Gly	Tyr	Asp	Gly	Gln	Asp	His	Leu	Glu	Phe	
		135					140					145				
TGC	CCT	GAC	ACA	CTG	GAT	TGG	AGA	GCA	GCA	GAA	CCC	AGG	GCC	TGG	CCC	713
Cys	Pro	Asp	Thr	Leu	Asp	Trp	Arg	Ala	Ala	Glu	Pro	Arg	Ala	Trp	Pro	
	150					155					160					
ACC	AAG	CTG	GAG	TGG	GAA	AGG	CAC	AAG	ATT	CGG	GCC	AGG	CAG	AAC	AGG	761
Thr	Lys	Leu	Glu	Trp	Glu	Arg	His	Lys	Ile	Arg	Ala	Arg	Gln	Asn	Arg	
	165				170					175					180	
GCC	TAC	CTG	GAG	AGG	GAC	TGC	CCT	GCA	CAG	CTG	CAG	CAG	TTG	CTG	GAG	809
Ala	Tyr	Leu	Glu	Arg	Asp	Cys	Pro	Ala	Gln	Leu	Gln	Gln	Leu	Leu	Glu	
				185					190					195		
CTG	GGG	AGA	GGT	GTT	TTG	GAC	CAA	CAA	GTG	CCT	CCT	TTG	GTG	AAG	GTG	857
Leu	Gly	Arg	Gly	Val	Leu	Asp	Gln	Gln	Val	Pro	Pro	Leu	Val	Lys	Val	
			200					205					210			
ACA	CAT	CAT	GTG	ACC	TCT	TCA	GTG	ACC	ACT	CTA	CGG	TGT	CGG	GCC	TTG	905
Thr	His	His	Val	Thr	Ser	Ser	Val	Thr	Thr	Leu	Arg	Cys	Arg	Ala	Leu	
		215					220					225				
AAC	TAC	TAC	CCC	CAG	AAC	ATC	ACC	ATG	AAG	TGG	CTG	AAG	GAT	AAG	CAG	953
Asn	Tyr	Tyr	Pro	Gln	Asn	Ile	Thr	Met	Lys	Trp	Leu	Lys	Asp	Lys	Gln	
	230					235					240					
CCA	ATG	GAT	GCC	AAG	GAG	TTC	GAA	CCT	AAA	GAC	GTA	TTG	CCC	AAT	GGG	1001
Pro	Met	Asp	Ala	Lys	Glu	Phe	Glu	Pro	Lys	Asp	Val	Leu	Pro	Asn	Gly	
	245				250					255					260	
GAT	GGG	ACC	TAC	CAG	GGC	TGG	ATA	ACC	TTG	GCT	GTA	CCC	CCT	GGG	GAA	1049
Asp	Gly	Thr	Tyr	Gln	Gly	Trp	Ile	Thr	Leu	Ala	Val	Pro	Pro	Gly	Glu	
				265				270						275		
GAG	CAG	AGA	TAT	ACG	TGC	CAG	GTG	GAG	CAC	CCA	GGC	CTG	GAT	CAG	CCC	1097
Glu	Gln	Arg	Tyr	Thr	Cys	Gln	Val	Glu	His	Pro	Gly	Leu	Asp	Gln	Pro	
			280					285					290			
CTC	ATT	GTG	ATC	TGG	GAG	CCC	TCA	CCG	TCT	GGC	ACC	CTA	GTC	ATT	GGA	1145
Leu	Ile	Val	Ile	Trp	Glu	Pro	Ser	Pro	Ser	Gly	Thr	Leu	Val	Ile	Gly	
		295					300					305				

GTC ATC AGT GGA ATT GCT GTT TTT GTC GTC ATC TTG TTC ATT GGA ATT	1193
Val Ile Ser Gly Ile Ala Val Phe Val Val Ile Leu Phe Ile Gly Ile	
310 315 320	
TTG TTC ATA ATA TTA AGG AAG AGG CAG GGT TCA AGA GGA GCC ATG GGG	1241
Leu Phe Ile Ile Leu Arg Lys Arg Gln Gly Ser Arg Gly Ala Met Gly	
325 330 335 340	
CAC TAC GTC TTA GCT GAA CGT GAG TGACACGCAG CCTGCAGACT CACTGTGGGA	1295
His Tyr Val Leu Ala Glu Arg Glu	
345	
AGGAGACAAA ACTAGAGACT CAAAGAGGGA GTGCATTTAT GAGCTCTTCA TGTTCAGGA	1355
GAGAGTTGAA CCTAAACATA GAAATTGCCT GACGAACTCC TTGATTTTAG CCTTCTCTGT	1415
TCATTTCTC AAAAAAGATTT CCCC	1440

(2) INFORMATION FOR SEQ ID NO:12:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 1440 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: cDNA

(ix) FEATURE:

- (A) NAME/KEY: CDS
- (B) LOCATION: 222..1268

(ix) FEATURE:

- (A) NAME/KEY: allele
- (B) LOCATION: replace(408, "g")
- (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis (HH)"
/label= 24d2

(ix) FEATURE:

- (A) NAME/KEY: allele
- (B) LOCATION: replace(1066, "a")
- (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis (HH)"
/label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:12:

GGGGACACTG GATCACCTAG TGTTTCACAA GCAGGTACCT TCTGCTGTAG GAGAGAGAGA	60
ACTAAAGTTC TGAAAGACCT GTTGCTTTTC ACCAGGAAGT TTTACTGGGC ATCTCCTGAG	120
CCTAGGCAAT AGCTGTAGGG TGA CTCTGG AGCCATCCCC GTTTCCCCGC CCCCCAAAAG	180
AAGCGGAGAT TTAACGGGGA CGTGCGGCCA GAGCTGGGGA A ATG GGC CCG CGA	233
Met Gly Pro Arg	

GCC Ala	AGG Arg	CCG Pro	GCG Ala	CTT Leu	CTC Leu	CTC Leu	CTG Leu	ATG Met	CTT Leu	TTG Leu	CAG Gln	ACC Thr	GCG Ala	GTC Val	CTG Leu	281
5					10					15					20	
CAG Gln	GGG Gly	CGC Arg	TTG Leu	CTG Leu	CGT Arg	TCA Ser	CAC His	TCT Ser	CTG Leu	CAC His	TAC Tyr	CTC Leu	TTC Phe	ATG Met	GGT Gly	329
				25					30					35		
GCC Ala	TCA Ser	GAG Glu	CAG Gln	GAC Asp	CTT Leu	GGT Gly	CTT Leu	TCC Ser	TTG Leu	TTT Phe	GAA Glu	GCT Ala	TTG Leu	GGC Gly	TAC Tyr	377
			40					45					50			
GTG Val	GAT Asp	GAC Asp	CAG Gln	CTG Leu	TTC Phe	GTG Val	TTC Phe	TAT Tyr	GAT Asp	GAT Asp	GAG Glu	AGT Ser	CGC Arg	CGT Arg	GTG Val	425
	55						60					65				
GAG Glu	CCC Pro	CGA Arg	ACT Thr	CCA Pro	TGG Trp	GTT Val	TCC Ser	AGT Ser	AGA Arg	ATT Ile	TCA Ser	AGC Ser	CAG Gln	ATG Met	TGG Trp	473
	70					75					80					
CTG Leu	CAG Gln	CTG Leu	AGT Ser	CAG Gln	AGT Ser	CTG Leu	AAA Lys	GGG Gly	TGG Trp	GAT Asp	CAC His	ATG Met	TTC Phe	ACT Thr	GTT Val	521
	85				90					95					100	
GAC Asp	TTC Phe	TGG Trp	ACT Thr	ATT Ile	ATG Met	GAA Glu	AAT Asn	CAC His	AAC Asn	CAC His	AGC Ser	AAG Lys	GAG Glu	TCC Ser	CAC His	569
				105					110				115			
ACC Thr	CTG Leu	CAG Gln	GTC Val	ATC Ile	CTG Leu	GGC Gly	TGT Cys	GAA Glu	ATG Met	CAA Gln	GAA Glu	GAC Asp	AAC Asn	AGT Ser	ACC Thr	617
			120					125					130			
GAG Glu	GGC Gly	TAC Tyr	TGG Trp	AAG Lys	TAC Tyr	GGG Gly	TAT Tyr	GAT Asp	GGG Gly	CAG Gln	GAC Asp	CAC His	CTT Leu	GAA Glu	TTC Phe	665
		135					140					145				
TGC Cys	CCT Pro	GAC Asp	ACA Thr	CTG Leu	GAT Asp	TGG Trp	AGA Arg	GCA Ala	GCA Ala	GAA Glu	CCC Pro	AGG Arg	GCC Ala	TGG Trp	CCC Pro	713
	150					155					160					
ACC Thr	AAG Lys	CTG Leu	GAG Glu	TGG Trp	GAA Glu	AGG Arg	CAC His	AAG Lys	ATT Ile	CGG Arg	GCC Ala	AGG Arg	CAG Gln	AAC Asn	AGG Arg	761
	165				170					175					180	
GCC Ala	TAC Tyr	CTG Leu	GAG Glu	AGG Arg	GAC Asp	TGC Cys	CCT Pro	GCA Ala	CAG Gln	CTG Leu	CAG Gln	CAG Gln	TTG Leu	CTG Leu	GAG Glu	809
				185					190					195		
CTG Leu	GGG Gly	AGA Arg	GGT Gly	GTT Val	TTG Leu	GAC Asp	CAA Gln	CAA Gln	GTG Val	CCT Pro	CCT Pro	TTG Leu	GTG Val	AAG Lys	GTG Val	857
			200					205					210			
ACA Thr	CAT His	CAT His	GTG Val	ACC Thr	TCT Ser	TCA Ser	GTG Val	ACC Thr	ACT Thr	CTA Leu	CGG Arg	TGT Cys	CGG Arg	GCC Ala	TTG Leu	905
			215				220					225				
AAC Asn	TAC Tyr	TAC Tyr	CCC Pro	CAG Gln	AAC Asn	ATC Ile	ACC Thr	ATG Met	AAG Lys	TGG Trp	CTG Leu	AAG Lys	GAT Asp	AAG Lys	CAG Gln	953
	230					235					240					
CCA Pro	ATG Met	GAT Asp	GCC Ala	AAG Lys	GAG Glu	TTC Phe	GAA Glu	CCT Pro	AAA Lys	GAC Asp	GTA Val	TTG Leu	CCC Pro	AAT Asn	GGG Gly	1001

245	250	255	260	
GAT GGG ACC TAC CAG GGC TGG ATA ACC TTG GCT GTA CCC CCT GGG GAA				1049
Asp Gly Thr Tyr Gln Gly Trp Ile Thr Leu Ala Val Pro Pro Gly Glu	265	270	275	
GAG CAG AGA TAT ACG TAC CAG GTG GAG CAC CCA GGC CTG GAT CAG CCC				1097
Glu Gln Arg Tyr Thr Tyr Gln Val Glu His Pro Gly Leu Asp Gln Pro	280	285	290	
CTC ATT GTG ATC TGG GAG CCC TCA CCG TCT GGC ACC CTA GTC ATT GGA				1145
Leu Ile Val Ile Trp Glu Pro Ser Pro Ser Gly Thr Leu Val Ile Gly	295	300	305	
GTC ATC AGT GGA ATT GCT GTT TTT GTC GTC ATC TTG TTC ATT GGA ATT				1193
Val Ile Ser Gly Ile Ala Val Phe Val Val Ile Leu Phe Ile Gly Ile	310	315	320	
TTG TTC ATA ATA TTA AGG AAG AGG CAG GGT TCA AGA GGA GCC ATG GGG				1241
Leu Phe Ile Ile Leu Arg Lys Arg Gln Gly Ser Arg Gly Ala Met Gly	325	330	335	340
CAC TAC GTC TTA GCT GAA CGT GAG TGACACGCAG CCTGCAGACT CACTGTGGGA				1295
His Tyr Val Leu Ala Glu Arg Glu	345			
AGGAGACAAA ACTAGAGACT CAAAGAGGGA GTGCATTTAT GAGCTCTTCA TGTTTCAGGA				1355
GAGAGTTGAA CCTAAACATA GAAATTGCCT GACGAACTCC TTGATTTTAG CCTTCTCTGT				1415
TCATTTCTCTC AAAAAGATTT CCCCA				1440

(2) INFORMATION FOR SEQ ID NO:13:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:13:

TGGCAAGGGT AAACAGATCC

20

(2) INFORMATION FOR SEQ ID NO:14:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:14:

CTCAGGCACT CCTCTCAACC

20

(2) INFORMATION FOR SEQ ID NO:15:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(ix) FEATURE:

- (A) NAME/KEY: modified_base
- (B) LOCATION: 1
- (D) OTHER INFORMATION: /mod_base= OTHER
/note= "N = 5'-biotinylated guanine
(bio-G) "

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:15:

NGAAGAGCAG AGATATACGT G

21

(2) INFORMATION FOR SEQ ID NO:16:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(ix) FEATURE:

- (A) NAME/KEY: modified_base
- (B) LOCATION: 1
- (D) OTHER INFORMATION: /mod_base= OTHER
/note= "N = 5'-biotinylated guanine
(bio-G) "

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:16:

NGAAGAGCAG AGATATACGT A

21

(2) INFORMATION FOR SEQ ID NO:17:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 18 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

- (ix) FEATURE:
 (A) NAME/KEY: modified_base
 (B) LOCATION: 1
 (D) OTHER INFORMATION: /mod_base= OTHER
 /note= "N = 5'-phosphorylated cytosine
 (p-C)"

- (ix) FEATURE:
 (A) NAME/KEY: modified_base
 (B) LOCATION: 18
 (D) OTHER INFORMATION: /mod_base= OTHER
 /note= "N = 3'-digoxigenin-conjugated
 guanine (G-dig)"

- (xi) SEQUENCE DESCRIPTION: SEQ ID NO:17:

NCAGGTGGAG CACCCAGN

18

- (2) INFORMATION FOR SEQ ID NO:18:

- (i) SEQUENCE CHARACTERISTICS:
 (A) LENGTH: 20 base pairs
 (B) TYPE: nucleic acid
 (C) STRANDEDNESS: single
 (D) TOPOLOGY: linear

- (ii) MOLECULE TYPE: DNA

- (xi) SEQUENCE DESCRIPTION: SEQ ID NO:18:

CTGAAAGGGT GGGATCACAT

20

- (2) INFORMATION FOR SEQ ID NO:19:

- (i) SEQUENCE CHARACTERISTICS:
 (A) LENGTH: 20 base pairs
 (B) TYPE: nucleic acid
 (C) STRANDEDNESS: single
 (D) TOPOLOGY: linear

- (ii) MOLECULE TYPE: DNA

- (xi) SEQUENCE DESCRIPTION: SEQ ID NO:19:

CAAGGAGTTC GTCAGGCAAT

20

- (2) INFORMATION FOR SEQ ID NO:20:

- (i) SEQUENCE CHARACTERISTICS:
 (A) LENGTH: 517 base pairs
 (B) TYPE: nucleic acid
 (C) STRANDEDNESS: single
 (D) TOPOLOGY: linear

- (ii) MOLECULE TYPE: DNA (genomic)

(ix) FEATURE:

- (A) NAME/KEY: -
- (B) LOCATION: 1..517
- (D) OTHER INFORMATION: /note= "normal or wild-type (unaffected) genomic sequence surrounding variant for 24d1(G) allele corresponding to positions 5507-6023 of genomic sequence containing the HH gene (SEQ ID NO:1)"

(ix) FEATURE:

- (A) NAME/KEY: allele
- (B) LOCATION: replace(328, "g")
- (D) OTHER INFORMATION: /phenotype= "normal or wild-type (unaffected)"
/label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:20:

```

TATTTCTTCTC CTCCAACCTA TAGAAGGAAG TGAAAGTTCC AGTCTTCCTG GCAAGGGTAA      60
ACAGATCCCC TCTCCTCATC CTTCTCTCTT CCTGTCAAGT GCCTCCTTTG GTGAAGGTGA      120
CACATCATGT GACCTCTTCA GTGACCACTC TACGGTGTCTG GGCCTTGAAC TACTACCCCC      180
AGAACATCAC CATGAAGTGG CTGAAGGATA AGCAGCCAAT GGATGCCAAG GAGTTCGAAC      240
CTAAAGACGT ATTGCCCAAT GGGGATGGGA CCTACCAGGG CTGGATAACC TTGGCTGTAC      300
CCCCTGGGGA AGAGCAGAGA TATACGTGCC AGGTGGAGCA CCCAGGCCTG GATCAGCCCC      360
TCATTGTGAT CTGGGGTATG TGAAGTATGA GAGCCAGGAG CTGAGAAAAT CTATTGGGGG      420
TTGAGAGGAG TGCCTGAGGA GGTAATTATG GCAGTGAGAT GAGGATCTGC TCTTTGTTAG      480
GGGGTGGGCT GAGGGTGGCA ATCAAAGGCT TTAAGTCTT                                517

```

(2) INFORMATION FOR SEQ ID NO:21:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 517 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(ix) FEATURE:

- (A) NAME/KEY: -
- (B) LOCATION: 1..517
- (D) OTHER INFORMATION: /note= "genomic sequence surrounding variant for 24d1(A) allele corresponding to positions 5507-6023 of genomic sequence containing the HH gene (SEQ ID NO:3)"

(ix) FEATURE:

- (A) NAME/KEY: allele
- (B) LOCATION: replace(328, "a")
- (D) OTHER INFORMATION: /phenotype= "Hereditary Hemochromatosis"

(HH) "
/label= 24d1

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:21:

TATTTTCCTTC CTCCAACCTA TAGAAGGAAG TGAAAGTTCC AGTCTTCCTG GCAAGGGTAA	60
ACAGATCCCC TCTCCTCATC CTTCTCTTTT CCTGTCAAGT GCCTCCTTTG GTGAAGGTGA	120
CACATCATGT GACCTCTTCA GTGACCACTC TACGGTGTCTG GGCCTTGAAC TACTACCCCC	180
AGAACATCAC CATGAAGTGG CTGAAGGATA AGCAGCCAAT GGATGCCAAG GAGTTCGAAC	240
CTAAAGACGT ATTGCCCAAT GGGGATGGGA CCTACCAGGG CTGGATAACC TTGGCTGTAC	300
CCCCTGGGGA AGAGCAGAGA TATACGTACC AGGTGGAGCA CCCAGGCCTG GATCAGCCCC	360
TCATTGTGAT CTGGGGTATG TGAAGTATGA GAGCCAGGAG CTGAGAAAAT CTATTGGGGG	420
TTGAGAGGAG TGCCTGAGGA GGTAATTATG GCAGTGAGAT GAGGATCTGC TCTTTGTTAG	480
GGGGTGGGCT GAGGGTGGCA ATCAAAGGCT TTAACCT	517

(2) INFORMATION FOR SEQ ID NO:22:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 361 amino acids
- (B) TYPE: amino acid
- (C) STRANDEDNESS:
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: protein

(ix) FEATURE:

- (A) NAME/KEY: Protein
- (B) LOCATION: 1..361
- (D) OTHER INFORMATION: /note= "Rabbit leukocyte antigen (RLA)"

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:22:

Met	Gly	Ser	Ile	Pro	Pro	Arg	Thr	Leu	Leu	Leu	Leu	Leu	Ala	Gly	Ala	1	5	10	15
Leu	Thr	Leu	Lys	Asp	Thr	Gln	Ala	Gly	Ser	His	Ser	Met	Arg	Tyr	Phe	20	25	30	
Tyr	Thr	Ser	Val	Ser	Arg	Pro	Gly	Leu	Gly	Glu	Pro	Arg	Phe	Ile	Ile	35	40	45	
Val	Gly	Tyr	Val	Asp	Asp	Thr	Gln	Phe	Val	Arg	Phe	Asp	Ser	Asp	Ala	50	55	60	
Ala	Ser	Pro	Arg	Met	Glu	Gln	Arg	Ala	Pro	Trp	Met	Gly	Gln	Val	Glu	65	70	75	80
Pro	Glu	Tyr	Trp	Asp	Gln	Gln	Thr	Gln	Ile	Ala	Lys	Asp	Thr	Ala	Gln	85	90	95	

[illegible]

(2) INFORMATION FOR SEQ ID NO:23:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 365 amino acids
(B) TYPE: amino acid
(C) STRANDEDNESS:
(D) TOPOLOGY: linear

(ii) MOLECULE TYPE: protein

(ix) FEATURE:

(A) NAME/KEY: Protein

(B) LOCATION: 1..365

(D) OTHER INFORMATION: /note= "Human Major Histocompatibility Class I (MHC) protein"

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:23:

```

Met Ala Val Met Ala Pro Arg Thr Leu Val Leu Leu Leu Ser Gly Ala
1          5          10          15

Leu Ala Leu Thr Gln Thr Trp Ala Gly Ser His Ser Met Arg Tyr Phe
          20          25          30

Phe Thr Ser Val Ser Arg Pro Gly Arg Gly Glu Pro Arg Phe Ile Ala
          35          40          45

Val Gly Tyr Val Asp Asp Thr Gln Phe Val Arg Phe Asp Ser Asp Ala
          50          55          60

Ala Ser Gln Arg Met Glu Pro Arg Ala Pro Trp Ile Glu Gln Glu Gly
65          70          75          80

Pro Glu Tyr Trp Asp Gly Glu Thr Arg Lys Val Lys Ala His Ser Gln
          85          90          95

Thr His Arg Val Asp Leu Gly Thr Leu Arg Gly Tyr Tyr Asn Gln Ser
          100          105          110

Glu Ala Gly Ser His Thr Leu Gln Met Met Phe Gly Cys Asp Val Gly
          115          120          125

Ser Asp Trp Arg Phe Leu Arg Gly Tyr His Gln Tyr Ala Tyr Asp Gly
          130          135          140

Lys Asp Tyr Ile Ala Leu Lys Glu Asp Leu Arg Ser Trp Thr Ala Ala
145          150          155          160

Asp Met Ala Ala Gln Thr Thr Lys His Lys Trp Glu Ala Ala His Val
          165          170          175

Ala Glu Gln Leu Arg Ala Tyr Leu Glu Gly Thr Cys Val Glu Trp Leu
          180          185          190

Arg Arg Tyr Leu Glu Asn Gly Lys Glu Thr Leu Gln Arg Thr Asp Ala
          195          200          205

Pro Lys Thr His Met Thr His His Ala Val Ser Asp His Glu Ala Thr
          210          215          220

Leu Arg Cys Trp Ala Leu Ser Phe Tyr Pro Ala Glu Ile Thr Leu Thr
225          230          235          240

Trp Gln Arg Asp Gly Glu Asp Gln Thr Gln Asp Thr Glu Leu Val Glu
          245          250          255

Thr Arg Pro Ala Gly Asp Gly Thr Phe Gln Lys Trp Ala Ala Val Val
          260          265          270

Val Pro Ser Gly Gln Glu Gln Arg Tyr Thr Cys His Val Gln His Glu
          275          280          285

```

Gly	Leu	Pro	Lys	Pro	Leu	Thr	Leu	Arg	Trp	Glu	Pro	Ser	Ser	Gln	Pro
290						295					300				
Thr	Ile	Pro	Ile	Val	Gly	Ile	Ile	Ala	Gly	Leu	Val	Leu	Phe	Gly	Ala
305					310					315					320
Val	Ile	Thr	Gly	Ala	Val	Val	Ala	Ala	Val	Met	Trp	Arg	Arg	Lys	Ser
				325					330					335	
Ser	Asp	Arg	Lys	Gly	Gly	Ser	Tyr	Ser	Gln	Ala	Ala	Ser	Ser	Asp	Ser
			340					345					350		
Ala	Gln	Gly	Ser	Asp	Val	Ser	Leu	Thr	Ala	Cys	Lys	Val			
		355					360					365			

(2) INFORMATION FOR SEQ ID NO:24:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:24:

ACATGGTTAA GGCCTGTTGC

20

(2) INFORMATION FOR SEQ ID NO:25:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:25:

GCCACATCTG GCTTGAAATT

20

(2) INFORMATION FOR SEQ ID NO:26:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(ix) FEATURE:

- (A) NAME/KEY: modified_base
- (B) LOCATION: 1

(D) OTHER INFORMATION: /mod_base= OTHER
 /note= "N = 5'-biotinylated adenine
 (bio-A) "

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:26:

NGCTGTTTCGT GTTCTATGAT C

21

(2) INFORMATION FOR SEQ ID NO:27:

(i) SEQUENCE CHARACTERISTICS:
 (A) LENGTH: 21 base pairs
 (B) TYPE: nucleic acid
 (C) STRANDEDNESS: single
 (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(ix) FEATURE:

(A) NAME/KEY: modified_base
 (B) LOCATION: 1
 (D) OTHER INFORMATION: /mod_base= OTHER
 /note= "N = 5'-biotinylated adenine
 (bio-A) "

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:27:

NGCTGTTTCGT GTTCTATGAT G

21

(2) INFORMATION FOR SEQ ID NO:28:

(i) SEQUENCE CHARACTERISTICS:
 (A) LENGTH: 19 base pairs
 (B) TYPE: nucleic acid
 (C) STRANDEDNESS: single
 (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(ix) FEATURE:

(A) NAME/KEY: modified_base
 (B) LOCATION: 1
 (D) OTHER INFORMATION: /mod_base= OTHER
 /note= "N = 5'-phosphorylated adenine
 (p-A) "

(ix) FEATURE:

(A) NAME/KEY: modified_base
 (B) LOCATION: 19
 (D) OTHER INFORMATION: /mod_base= OTHER
 /note= "N = 3'-digoxigenin-conjugated
 adenine (A-dig) "

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:28:

NTGAGAGTCG CCGTGTGGN

19

(2) INFORMATION FOR SEQ ID NO:29:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 39 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:29:

GGAAGAGCAG AGATATACGT GCCAGGTGGA GCACCCAGG

39

(2) INFORMATION FOR SEQ ID NO:30:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 39 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:30:

GGAAGAGCAG AGATATACGT ACCAGGTGGA GCACCCAGG

39

(2) INFORMATION FOR SEQ ID NO:31:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 22 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:31:

CAAAAGAAGC GGAGATTTAA CG

22

(2) INFORMATION FOR SEQ ID NO:32:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 19 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:32:

AGATTTAACG GGGACGTGC

19

(2) INFORMATION FOR SEQ ID NO:33:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 22 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:33:

AGAGGTCACA TGATGTGTCA CC

22

(2) INFORMATION FOR SEQ ID NO:34:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 19 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:34:

AGGAGGCACT TGTGTGTCC

19

(2) INFORMATION FOR SEQ ID NO:35:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 21 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:35:

AAAATCACAA CCACAGCAAA G

21

(2) INFORMATION FOR SEQ ID NO:36:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:36:

TTCCCACAGT GAGTCTGCAG

20

(2) INFORMATION FOR SEQ ID NO:37:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 19 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:37:

CAATGGGGAT GGGACCTAC

19

(2) INFORMATION FOR SEQ ID NO:38:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:38:

ATATACGTGC CAGGTGGAGC

20

(2) INFORMATION FOR SEQ ID NO:39:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 20 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:39:

CCTCTTCACA ACCCCTTTCA

20

(2) INFORMATION FOR SEQ ID NO:40:

- (i) SEQUENCE CHARACTERISTICS:
 - (A) LENGTH: 22 base pairs
 - (B) TYPE: nucleic acid
 - (C) STRANDEDNESS: single
 - (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:40:

CATAGCTGTG CAACTCACAT CA

22

(2) INFORMATION FOR SEQ ID NO:41:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 40 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:41:

AGCTGTTCGT GTTCTATGAT CATGAGAGTC GCCGTGTGGA

40

(2) INFORMATION FOR SEQ ID NO:42:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 40 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:42:

AGCTGTTCGT GTTCTATGAT GATGAGAGTC GCCGTGTGGA

40

(2) INFORMATION FOR SEQ ID NO:43:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 32 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single
- (D) TOPOLOGY: linear

(ii) MOLECULE TYPE: DNA (genomic)

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:43:

TGTTCTATGA TCATGAGAGT CGCCGTGTGG AG

32

(2) INFORMATION FOR SEQ ID NO:44:

(i) SEQUENCE CHARACTERISTICS:

- (A) LENGTH: 32 base pairs
- (B) TYPE: nucleic acid
- (C) STRANDEDNESS: single

(ii) MOLECULE TYPE: DNA (genomic)

(xi) SEQUENCE DESCRIPTION: SEQ ID NO:44:

TGTTCTATGA TCATGAGTGT CGCCGTGTGG AG